Türkiye Maternal Fetal Tıp ve Perinatoloji Derneği

Ultrasonografi Kongresi

Maternal - Fetal Tıp ve Perinatoloji Derneği Türkiye

23-06-1998

29 Ekim – 1 Kasım 2025

Renaissance Polat İstanbul Hotel
Yeşilköy

BİLDİRİ KİTABI

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DAVET

Değerli Meslektaşlarımız,

Türkiye Maternal Fetal Tıp ve Perinatoloji Derneği'nin geleneksel Ultrasonografi Kongresi 29 Ekim - 1 Kasım 2025 tarihleri arasında Renaissance Polat İstanbul Hotel Yeşilköy'de düzenlenecektir.

Kongre öncesinde, 29 Ekim 2025 tarihinde, ISUOG onaylı İleri Düzey Fetal Ultrasonografi Kursu gerçekleştirilecektir. Bu önemli eğitim programında fetal anatomi tarama protokolleri, doppler teknikleri ve fetal malformasyonların değerlendirilmesi gibi ileri düzey konular ele alınacaktır.

Bu dört günlük toplantıda, obstetrik ultrasonografi ile ilgili en güncel bilgiler interaktif şekilde ele alınacak, tanıya giden ipuçları, tuzaklar tartışılacaktır. Her zamanki gibi ülkemiz ve diğer ülkelerdeki meslektaşımızın deneyimlerinden yararlanma imkânı olacaktır.

Deneyimi olan herkesin katkısı ve genç meslektaşlarımızın ana salonda sunma olanağı bulacağı sözel bildiriler ve posterle zenginlik kazanacaktır. Ayrıca hasta başı uygulamalarıyla pratik konular ele alınacaktır.

Gebe takibiyle uğraşan bütün Kadın-Doğum Hekimleri, Perinatoloji Uzmanları ve ilgili diğer branşlardaki meslektaşlarımızın bilgi alışverişinde bulunma ve endüstri temsilcileriyle bir araya gelerek, prenatal tanı yöntemleri, genetik ve ultrasonografi teknolojisindeki en son yenilikleri görme şansına sahip olacağını bilmenizi isteriz.

Katılan herkes için yararlı olacağına samimiyetle inandığımız bu bilimsel şölene sizleri en içten dileklerimizle davet ediyoruz.

Prof. Dr. Rıza MADAZLI
Dernek ve Kongre Başkanı

Prof. Dr. Recep HAS
Kongre Başkanı

KURULLAR

ORGANİZASYON KURULU

KONGRE BAŞKANLARI
Rıza MADAZLI
Recep HAS

KONGRE SEKRETERI
İnanç MENDİLCİOĞLU

BILIMSEL KURUL
(Türkiye Maternal Fetal Tıp ve Perinatoloji Derneği Yönetim Kurulu)

Rıza MADAZLI
Recep HAS
İnanç MENDİLCİOĞLU
Şevki ÇELEN
İbrahim KALELİOĞLU
Acar KOÇ
Sermet SAĞOL
Filiz YANIK
Gökhan YILDIRIM

BILIMSEL PROGRAM

SALON A	29 EKIM 2025
08.50-09.00	**Açılış** Prof. Dr. Recep HAS, *Kongre Başkanı* Prof. Dr. Inanc MENDILCIOGLU, *Dernek ve Kongre Genel Sekreteri*
09.00-10.30	**1. Oturum** **Oturum Başkanları: Recep Has, İnanç Mendilcioğlu**
09.00-09.20	**Ventrikülomegali** Atıl Yüksel
09.20-09.50	**Korpus kallozum anomalileri** Dario Paladini
09.50-10.10	**SSS anomalilerinde şüpheli bulgular, ipuçları ve tuzaklar** Gustavo Malinger (çevrimiçi)
10.10-10.30	**Canlı ultrason: İlk trimester SSS ultrasonu** Dario Paladini
10.30-11.00	**Kahve Molasi**
11.00-13.10	**2. Oturum** **Oturum Başkanları: Rıza Madazlı, Özgür Deren**
11.00-11.30	**İlk trimesterde SSS: Ne görebiliriz?** Reem S. Abu-Rustum
11.30-12.00	**Fetal NSG patern tanımlama ve prenatal genetik tanı yaklaşımı** Ritsuko Pooh
12.00-12.30	**Görüntüye hakim olmak: TV-NSG için optimal planlarda pratik ipuçları** Ritsuko Pooh
12.30-13.10	**Canlı ultrason: Transabdominal – Transvajinal SSS ultrasonu** Ritsuko Pooh
13.10-14.00	**Öğle yemeği**

14.00-16.20	**3. Oturum** **Oturum Başkanları: Acar Koç, Fehmi Yazıcıoğlu**
14.00-14.30	**Dört boşluk görünümü: İpuçları ve püf noktaları** Reem S. Abu-Rustum
14.30-14.50	**Atabiner Aortanın Ayırıcı Tanısı** Yalçın Kimya
14.50-15.10	**Kardiyak ventrikül disproporsiyonu** Dario Paladini
15.10-15.40	**Fetal kalpte normal ve anormal venöz bağlantılar** Alfred Abuhamad (çevrimiçi)
15.40-16.20	**Canlı ultrason: İlk ve ikinci trimester kardiyak ultrason** Reem S. Abu-Rustum
16.20-16.50	**Kahve Molasi**
16.50-18.40	**4. Oturum** **Oturum Başkanları: Nuri Danışman, Tamer Mungan**
16.50-17.10	**Preeklampsi öngörüsünde Doppler kullanımı** Liona Poon
17.10-17.30	**Aortik ark anomalileri** Halil Aslan
17.30-17.50	**Fetal kalp ritim bozuklukları** Sudhir Vashist (çevrimiçi)
17.50-18.20	**Canlı ultrason: Obstetrik Doppler** Liona Poon
18.20-18.40	**Kapanış**

SALON A	30 EKIM 2025
08.00-08.20	**AÇILIŞ** Prof. Dr. Rıza MADAZLI, *Dernek Başkanı* Prof. Dr. Recep HAS, *Kongre Başkanı* Prof. Dr. Inanc MENDILCIOGLU, *Dernek ve Kongre Genel Sekreteri*
08.20-09.40	**1.OTURUM: MERKEZİ SİNİR SİSTEMİ** **Oturum Başkanları:** Sinan Beksaç, Namık Demir
08.20-08.30	**SS-001 İlk trimesterde ventrikülotalamik açının fetal büyüme kısıtlılığının yeni bir öngörücüsü olarak değerlendirilmesi: prospektif kohort çalışma** Yasemin Dadaş
08.30-08.50	**Anterior kompleksin nörosonografik değerlendirilmesi** Fernando Vinals (online)
08.50-09.10	**Kortikal malformasyonların tanısında ipuçları** Ritsuko Pooh
09.10-09.30	**Orta ve arka beyin anormallikleri** Ritsuko Pooh
09.30-09.40	**Tartışma**
09.40-10.10	**UYDU SEMPOZYUMU-1: PFIZER**
09.40-10.10	**Abrysvo ile RSV'ye karşı maternal immunizasyon: İlk nefesten itibaren koruma** Acar Koç, Merih Çetinkaya
10.10-10.40	**Kahve Molası**
10.40-11.20	**ANA KONUŞMA-1** **Moderatör:** Nuri Danışman, Feride Söylemez
10.40-10.55	**Temel ultrasonografi eğitimi** Reem S. Abu-Rustum
10.55-11.10	**Sonopartogram** Liona Poon
11.10-11.20	**Tartışma**

11.20-13.10	**FETAL ANOMALİLER PANELİ-1: MERKEZİ SİNİR SİSTEMİ ANOMALİLERİ / VAKA SUNUMLARI / SÖZEL BİLDİRİLER- 2 ve SORU-CEVAPLAR**
11.20-11.40	**MSS (CANLI USG-1)** Ritsuko Pooh
	Panel Başkanları: Atıl Yüksel, Esra Esim Büyükbayrak, Zeki Şahinoğlu
	Panelistler: Bilge Çetinkaya Demir, Aytül Çorbacıoğlu Esmer, Tuğba Saraç Sivrikoz, Hakan Erenel, Ali Ekiz, Süreyya Sarıdaş Demir, Erkan Çağlıyan, Selen Gürsoy Erzincan, Çiğdem Kunt İşgüder
	- Lobar Holoprosensefalide “snake under skull sign” bulgusu, **Ali Ekiz** - İzole septum pellusidum agenezisi, **Çiğdem Kunt İşgüder** - Ventrikülomegali, **Aytül Çorbacıoğlu** - Blake poş kisti, **Selen Gürsoy Erzincan** - CMV enfeksiyonunda prenatal ultrason bulguları, **Hakan Erenel** - Araknoid kist-retroorbital tümör olgusu, **Tuğba Saraç Sivrikoz** - İntrakraniyal malign tümör, **Bilge Çetinkaya** - Dural sinüs AV malformasyonu, **Erkan Çağlıyan** - Galen veni anevrizması, **Süreyya Sarıdaş Demir** - Açık spina bifidada fetal cerrahi, **Esra Esim Büyükbayrak**
	SS-002 Prenatal tanı konan izole korpus kallosum anomalilerinde nörogelişimsel sonuçlar Cem Dağdelen
13.10-14.00	**Öğle Yemeği**
14.00-15.40	**2. OTURUM: PREEKLAMPSİ, DOPPLER, ERKEN DOĞUM** **Oturum Başkanları:** Şevki Çelen, İskender Başer
14.00-14.20	**Doppler USG (CANLI USG-2)** Liona Poon
14.20-14.30	**SS-003 Sezaryen sırasında uterus nasıl kapatılmalı? Çevrimiçi anket çalışması** Gürcan Türkyılmaz
14.30-14.50	**Preeklampsinin erken prediksiyonu** Liona Poon
14.50-15.10	**Preeklampsi yönetiminde anjiogenik belirteçler** Liona Poon
15.10-15.30	**Preterm doğumun önlenmesi** Ozhan Turan (online)
15.30-15.40	**Tartışma**

15.40-16.25	**UYDU SEMPOZYUMU-2 & CANLI USG DEMO: PROMEDIS (SAMSUNG)** **Moderatör:** Rıza Madazlı
15.40-16.25	**İkinci trimester ultrason taramasında eş zamanlı yapay zeka desteği** Daniele Di Mascio
16.25-16.55	**UYDU SEMPOZYUMU-4: NATERA** **Moderatör:** Acar Koç
16.25-16.55	**NIPT'de pratiği değiştiren yenilikler ve gebelik planlaması ve takibinde taşıyıcılık tarama testlerinin önemi** Samantha Leonard
16.55-18.25	**FETAL ANOMALİLER PANELİ-2: BİRİNCİ TRİMESTER ANOMALİLER / GASTROİNTESTİNAL ANOMALİLER / VAKA SUNUMLARI / SÖZEL BİLDİRİ-4 ve SORU-CEVAPLAR**
	Panel Başkanları: Sermet Sağol, Mustafa Başbuğ, Gökhan Yıldırım
	Panelistler: Aybike Pekin, Yasemin Doğan, Alev Atış Aydın, Mehmet Osmanağaoğlu, Umutcan Kayıkçı, Hülya Dede, Korkut Dağlar, Halis Özdemir
	- Kistik higroma, **Yasemin Doğan** - Ensefalosel ve polikistik böbrek (Meckel Gruber sendromu), **Aybike Pekin** - Kardiyak anomali, **Hülya Dede** - Triküspit atrezisi, **Umutcan Kayıkçı** - Omfalosel, **Alev Atış Aydın** - Trakeoözofagial fistül ile birlikte olan multipl anomali vakası, **Halil Özdemir** - Özofagus atrezisi ve/ veya Özofagus atrezisi + Trakeoözofagial fistül olan 12 vakanın sunumu, **Mehmet Osmanağaoğlu** - İzole Anal Atrezi, **Korkut Dağlar**
	SS-004 Fetal intraabdominal kalsifikasyonlu gebeliklerin prenatal ve postnatal takibi: tek merkez deneyimi Elif Ayan Avcı

18.25-19.55	**FETAL ANOMALİLER PANELİ-3: TORAKS ANOMALİLERİ / İSKELET DİSPLAZİLERİ / VAKA SUNUMLARI / SÖZEL BİLDİRİ-5 ve SORU-CEVAPLAR**
	Panel Başkanları: Tamer Mungan, Ali Ergün, Aydan Biri
	Panelistler: Murat Çağan, Başak Kaya, Nihal Şahin Uysal, Gökçen Örgül, Didem Kaymak, Zehra Vural, Semir Köse, Gürcan Türkyılmaz
	- El anomalileri, **Gürcan Türkyılmaz** - Geç başlangıçlı hidrotoraks, **Murat Çağan** - Fetal sırt ve üst mediasteni içine alan tümör, **Murat Çağan** - Diyafragma eventrasyonu, **Başak Kaya** - Konjenital pulmoner hava yolu malformasyonu, **Başak Kaya** - TRIP 11 gen mutasyonuna bağlı iskelet sistemi displazisi, **Semir Köse** - Akondrogenezis Tip 2 (Langer-Saldino), **Didem Kaymak** - Vertebral laminanın erken osifikasyonu, **Didem Kaymak** - Saethe Chotzen sendromu, **Nihal Şahin Uysal** - Mediastinal Teratom, **Nihal Şahin Uysal** - İzole plevral efüzyon olgusunda yaklaşım, **Zehra Vural** - Short rib polidaktili sendromu, **Zehra Vural** - Rubinstein–Taybi Sendromu, **Gökçen Örgül** - Plevral efüzyon ile komplike CCAM, **Gökçen Örgül**
	SS-005 Fetal yapısal kardiyak anomaliler dışındaki doğuştan kardiyak ve orta mediasten anomalilerinin perinatal sonuçları Aybike Kaya

SALON A	31 EKIM 2025
08.00-09.30	**3.OTURUM: ÇOĞUL GEBELİKLER-CMV** **Oturum Başkanları:** Mert Kazandı, Mehmet Şimşek
08.00-08.10	**SS-006 Nullipar kadınlarda aktif doğum eyleminin başlangıcında intrapartum ultrason kullanılarak doğum zamanının öngörülmesi** Murat Aykut Cantürk
08.10-08.20	**SS-007 Term gebeliklerde doğum eylemini başlatma/indüklemede meme stimülasyonunun maternal ve fetal sonuçları** Ayşe Sanem Öksüz
08.20-08.30	**SS-008 Gebelikte madde kullanımının maternal ve neonatal sonuçları: retrospektif kohort** Nurten Cilek
08.30-08.45	**1.Trimester çoğul gebelik ultrasonografisi** Oya Demirci
08.45-09.05	**Konjenital CMV önlenmesi ve yönetimi** Daniele Di Mascio
09.05-09.25	**İkiz gebeliklerde büyüme değerlendirmesi** Daniele Di Mascio
09.25-09.30	**Tartışma**
09.30-10.00	**UYDU SEMPOZYUMU-3: SANOFI** **Moderatör:** Filiz Yanık
09.30-10.00	**Boğmaca ile mücadelede stratejik adım: Multidispliner yaklaşımla ulusal Tdab uygulaması** Özlem Pata, Dilek Yılmaz
10.00-10.30	**Kahve Molası**

10.30-12.20	**FETAL ANOMALİLER PANELİ-4: ÇOĞUL GEBELİKLER / FETAL BÜYÜME KISITLILIĞI / VAKA SUNUMLARI / SÖZEL BİLDİRİLER 9 ve SORU-CEVAPLAR**
10.30-10.50	**2. Trimester İkiz USG (CANLI USG-3)** Daniele Di Mascio
	Panel Başkanları: Babür Kaleli, İbrahim Kalelioğlu, Fuat Akercan
	Panelistler: Aytaç Yüksel, Gül Alkan Bülbül, Melih Velipaşaoğlu, Umut Dilek, Doruk Cevdi Katlan, Ayşegül Özel, Banu Dane, Lütfiye Uygur
	- Koryonisite, **Aytaç Yüksel** - Embriyo redüksiyonu, **Aytaç Yüksel** - Selektif redüksiyon teknikleri, **Gül Alkan Bülbül** - Dikoryoniklerde antenatal bakım, **Gül Alkan Bülbül** - Monokoryoniklerde antenatal bakım, doğum zamanlaması, **Melih Velipaşaoğlu** - Monoamniyotiklerde antenatal bakım, doğum zamanlaması, **Melih Velipaşaoğlu** - Monokoryoniklerde takibi gereken parametreler, **Umut Dilek** - Monokoryoniklerde nörolojik sekel nedenleri, **Umut Dilek** - Monokoryoniklerde nörolojik etkilenim bulguları: US ve MR, **Doruk Cevdi Katlan** - Kombine test ve CffDNA: Genetik zigozite, **Doruk Cevdi Katlan** - İkizlerde CVS mi AS mi? Tek örnek çift örnek, **Ayşegül Özel** - Ikizlerde kısa kollum yönetimi, progesteron ve serklaj, **Ayşegül Özel** - Ikizlerde preeklampsi öngörüsü aspirin ile korunma, **Banu Dane** - TTTS TAPS Diskordan gelişim: Tanı, sınıflama, yönetim, **Banu Dane** - Akardiyak ikiz ve yapışık ikizler: Tanı yönetim, **Lütfiye Uygur** - MK ve DK'larda diskordan gelişimde doğum zamanlaması, **Lütfiye Uygur**
	SS-009 İleri evre ikiz anemi-polisitemi sekansı (TAPS) vakalarına iki farklı yaklaşım Tuğçe Tunç Acar
12.20-13.10	**Öğle Yemeği**
13.10-13.30	**ANA KONUŞMA-2** **Moderatör:** Recep Has, Mehmet Uludoğan
13.10-13.30	**Antenatal bakımda güncel gelişmeler** Kypros Nicolaides (online)

13.30-15.00	**4.OTURUM: FETAL KALP-YÜZ** **Oturum Başkanları:** İnanç Mendilcioğlu, Tuncay Nas
13.30-13.50	**Kalp (CANLI USG-4)** Dario Paladini
13.50-14.10	**1. Timester kalp değerlendirmesi** Reem S. Abu-Rustum
14.10-14.30	**Kardiyak septal defektler** Dario Paladini
14.30-14.50	**Mikrognati** Dario Paladini
14.50-15.00	**Tartışma**
15.00-15.30	**Kahve Molası**
15.30-16.00	**UYDU SEMPOZYUMU-5: GEN-ERA**
15.30-16.00	**Prenatal izlemlerde non-invaziv prenatal testin (NIPT) klinik önemi** Boutros Maroun
16.00-17.30	**FETAL ANOMALİLER PANELİ-5: KARDİAK ANOMALİLER, DOPPLER USG / VAKA SUNUMLARI / SÖZEL BİLDİRİLER-10 ve SORU-CEVAPLAR**
	Panel Başkanları: Özgür Deren, Halil Aslan, Özlem Pata
	Panelistler: Ebru Davutoğlu, Münip Akalın, Erdem Fadıloğlu, Reyhan Ayaz, İlker Ali Çerçi, Miraç Özalp, Işıl Ayhan, Serdar Kaya, Serdar Kütük
	- Atrioventriküler Septal Defekt (AVSD), **Ebru Davutoğlu** - Pulmoner Kapak Yokluğu Sendromu (APVS), **İlker Ali Çerçi** - Çift Çıkışlı Sağ Ventrikül (DORV-Retinoik asit embriyopatisi), **Işıl Ayhan** - Fetal Kardiak Girişimler, **Erdem Fadıloğlu** - Vena kava inferior anevrizması, **Miraç Özalp** - Heterotaksi Sendromu, **Münip Akalın** - Fetal Rabdomyoma, **Serdar Kaya** - Fetal Aritmi, **Reyhan Ayaz** - Portal sistem agenezisi, **Serdar Kütük**
	SS-010 Konjenital scimitar sendromu: prenatal tanı ve vaka sunumu Zinar Aydoğan

17.30-18.50	**5.OTURUM: OBSTETRİK KANAMALAR** **Oturum Başkanları:** Aykan Yücel, Alkan Yıldırım
17.30-17.45	**Sezaryen skar gebeliği** Özgür Özyüncü
17.45-18.05	**Kritik obstetrik hastabaşı USG** Allison Lankford (online)
18.05-18.25	**Plasental invazyon anomalilerinde sezaryen histerektomi-uterus koruyucu cerrahi prediksiyonu** Ahmed M. Hussein (online)
18.25-18.40	**Plasental invazyon anomalilerinde yönetim** Tuncay Özgün
18.40-18.50	**Tartışma**

SALON B	31 EKIM 2025
10.30-12.00	**SÖZEL BİLDİRİLER OTURUMU - 1** **Oturum Başkanları:** Ali Çetin, Füsun Varol
10.30-12.00	**SS-018 Fitobezoar ilişkili mekanik bağırsak tıkanıklığı ile seyreden geç preterm gebelik: vaka sunumu** Turgay Hakkı Emet **SS-019 Konjenital diyafragma hernisi: 10 yıllık tek merkez deneyimi** Gizem Pınar **SS-020 Monokoryonik diamniyotik ikiz gebelikte her iki fetüste izole barsak herniasyonu ile seyreden konjenital diyafragma hernisi: prenatal tanıda doppler ultrasonun rolü** Candan Din **SS-021 Ultrason rehberliğinde mikrodalga ablasyon ile tedavi edilen plasental koryoanjiyoma vakası** Figen Günday **SS-022 Prenatal dönemde saptanan umbilikal kord kistleri: klinik özellikleri ve perinatal sonuçları** Çağdaş Nurettin Emeklioğlu **SS-023 Eşzamanlı NSD1 ve WDR11 mutasyonları ile Sotos sendromunun prenatal tanısı: nadir bir vaka sunumu** Zehra Tavukçuoğlu **SS-024 Polimeli vakasının prenatal tanısı ve postnatal yönetimi: nadir vaka sunumu** Betül Tokgöz Çakır **SS-025 Spontan rezolüsyon gösteren fetal megalouretra prenatal tanısı: iki vaka sunumu** İsmail Yılmaz **SS-026 Fetal posterior üretral valv ile ilişkili fetal mesane rüptürüne bağlı nadir fetal asit vakası: vaka sunumu** Aybüke Fatma Dirgen Yakar **SS-027 Fetüsten babaya: prenatal CMT1A tanısının ailesel genetik taramaya yol açtığı nadir vaka** Hale Özer Çaltek **SS-028 Polihidramniyoz ile komplike olan üçüncü trimester fetal servikal teratom yönetimi için EXIT prosedürü: vaka sunumu** Engin Öztürk **SS-029 Ellis–van Creveld sendromu: vaka sunumu** Aslı Altınordu Atıcı **SS-030 Akraba evliliğinde çoklu konjenital anomalilerle birlikte görülen Ektrodaktili-Ektodermal Displazi-Yarık Dudak Sendromunun prenatal tespiti** Reşat Mısırlıoğlu **SS-031 Ölümcül osteogenezis imperfekta tip II** Mensure Tonguc **SS-032 Nadir bir toraks hipoplazisi nedeni olarak Raine sendromu** Coşkun Ümit **SS-033 Kalıcı fetal bradikardi için salbutamol kullanımı: vaka sunumu** Erkan Çağlıyan

13.30-15.00	**SÖZEL BİLDİRİLER OTURUMU - 2** **Oturum Başkanları:** İrfan Kutlar, Kazım Emre Karaşahin
13.30-15.00	**SS-034 32. gebelik haftasından önceki preterm doğumlarda antenatal magnezyum sülfat nöroproteksiyonunun farklı sürelerle ilişkili erken ve geç neonatal sonuçlarının değerlendirilmesi** Selman Alikiliç **SS-035 Doğum ağırlığının yenidoğan yoğun bakım yatışları ile ilişkisi: 1256 doğumun retrospektif analizi** Engin Öztürk **SS-036 İntrapartum ilerleme açısı ve baş-perine mesafesi ölçümünün doğum şekli ve süresini öngörmedeki rolünün değerlendirilmesi** Merve Balki Yüzüak **SS-037 Drenaj ve arteriyel embolizasyon ile yönetilen postpartum retroperitoneal hematom: iki vaka sunumu** Merve Keskin Paker **SS-038 Bilincini kaybeden term gebeye yaklaşım – hemorajik inme ve eklampsi yönetimi** Şule Birol İnce **SS-039 Fetal hepatosplenomegali ile prezente olan konjenital sifiliz vakası** Abdullah Tabakçı **SS-040 Maternal propiltiyourasil kullanımına bağlı intrauterin büyüme kısıtlılığı ile ilişkili fetal kalp yetmezliğinin sonografik gösterimi** Tuncay Karaahmetoglu **SS-041 Sınıf A1 gestasyonel diyabet tanısı almış gebelerde serum inflamatuar belirteçlerinin değerlendirilmesi** Ezgi Bilicen **SS-042 Postpartum retroperitoneal hematom: konservatif yaklaşımla yönetilen bir vaka** Kubilay Çanga **SS-043 Çoklu fetal anomalilerle birlikte Turner sendromunun prenatal tanısı** Meltem Çalışkan **SS-044 Wolfram sendromlu hastada başarılı gebelik: vaka sunumu** Esra Nuhoğlu Bilgin **SS-046 Nörofibromatozis tip 1'li bir hastada Emanuel sendromu: ardışık iki gebeliğin yönetimi** Ayşegül Ünal Gören **SS-047 Fetal koledokolitiazis tanısı konan 3 vakanın prenatal tanısı ve postnatal sonuçları: vaka serisi** Fatih Barutçu **SS-048 Postnatal olarak FANCF mutasyonu ve eşlik eden PEX7 varyantı ile tanı konan nadir prenatal multisistem anomalisi vakası** Fatma Nur Ceylan

SALON A	1 KASIM 2025
08.00-09.45	**6.OTURUM: BİRİNCİ TRİMESTER USG** **Oturum Başkanları:** Cüneyt Evrüke, Filiz Yanık
08.00-08.10	**SS-011 Homozigot KHDC3L varyantı taşıyıcısında tekrarlayan molar gebelik** Esra Selvi
08.10-08.20	**SS-012 Fetal intrakraniyal kanama tanısı alan olguların postnatal sonuçları** Cem Dağdelen
08.20-08.30	**SS-013 Poliarteritis Nodosa Ve Preeklampsi Zemininde Gelişen Renal Hematom:Vaka Sunumu** Tahir Kaçar
08.30-08.45	**Erken gebelikte normal anormal bulgular** Selim Büyükkurt
08.45-09.00	**Taşıyıcılık testleri kime ne zaman nasıl?** Hülya Kayserili
09.00-09.15	**Noninvazif prenatal tanıda yenilikler** Tuğba Kalaycı
09.15-09.35	**Birinci trimesterde tanı konamayan ilginç vakalar** Reem S. Abu-Rustum
09.35-09.45	**Tartışma**
09.45-10.15	**UYDU SEMPOZYUMU-5: EXELTIS**
09.45-10.15	**Regenesis Max: Annelik yolculuğunda güvenli formülasyon** Oluş Api
10.15-10.45	**Kahve Molası**
10.45-12.35	**FETAL ANOMALİLER PANELİ-6: cfDNA PANELİ: cfDNA TESTLERİ VE FETAL USG BULGULARI / VAKA SUNUMLARI / SÖZEL BİLDİRİLER-14 ve SORU-CEVAPLAR**

10.45-11.05	**1. Trimester USG (CANLI USG-5)** Reem S. Abu-Rustum
	Panel Başkanları: Acar Koç, Seher Başaran, Sabahattin Altınyurt
	Panelistler: Mert Turğal, Cihan İnan, Erzat Toprak, Deniz Karcaaltıncaba, Tuğba Kalaycı, Aslı Göker, Filiz Yanık, Ebru Çelik
	Konu Başlıkları: cfDNA kime, ne zaman ve nasıl? Sonuç verilemeyen cfDNA testi, mikrodelesyon-duplikasyonları ve cinsiyet kromozom anomalilerini tarayalım mı?, Tüm kromozomlar? USG bulguları normal/anormal ve cfDNA sonuçlarını nasıl değerlendirelim? Gebelik komplikasyonları ve cfDNA. Maternal sağlık, cell based NIPT, taşıyıcılık taramaları, WES, WGS.
	SS-014 Uyumsuz anormal NIPT ve/veya CVS sonuçları ile normal birinci trimester ultrason bulguları olan gebeliklerde takip ve perinatal sonuçlar: vaka serisi Ekrem Ergenç
12.35-14.00	**Öğle Yemeği**
14.00-15.30	**FETAL ANOMALİLER PANELİ-7: GENİTOÜRİNER ANOMALİLER, KRANİYOFASİYAL ANOMALİLER / VAKA SUNUMLARI / SÖZEL BİLDİRİLER-15 ve SORU-CEVAPLAR**
	Panel Başkanları: Cem Batukan, Cenk Sayın, Gülseren Yücesoy
	Panelistler: Cem Sanhal, Verda Alpay, Fedi Ercan, Oluş Api, Şükran Doğru, Derya Eroğlu, Ali Özgür Ersoy, Deniz Kanber Acar
	- Artmış NT, bilateral pelviektazi ve hidrops ile renal displazi gelişen CVS ile WES analizinde "Multiple Anomalies- Seizure -Hypotonia Syndrome" tanısı alan olgu, **Ali Özgür Ersoy** - Pierre Robin Sendromu olgusu (İzole damak yarığı, mikrognati), **Cem Sanhal** - Dil kökünde Ranula olgusu, **Fedi Ercan** - Boyunda branchial kist olgusu, **Fedi Ercan** - Megalouretra olgusu, **Verda Alpay** - Sirolimus ile tedaviye yanıt veren boyunda dev lenfanjiom olgusu, **Verda Alpay** - Ağız içi kitle (Epulis), **Şükran Doğru** - Kloakal ekstrofi), **Şükran Doğru** - Anal Atrezi, kloaka anomalisi olgusu, **Deniz Kamber Acar** - Amniotik banta bağlı anterior ensefalosel ve fasial cleft olgusu, **Deniz Kamber Acar** - Fetal Papillorenal Sendrom olgusu, **Derya Eroğlu** - Fetal pelvik böbrek olgusu, **Derya Eroğlu** - Tip1 Posterior uretral valv - olgusu (vezikoamniotik şant uygulaması), **Oluş Api**
	SS-015 Fetal renal füzyon anomalileri ve perinatal sonuçları Aybike Kaya
15.30-16.00	**Kahve Molası**

16.00-17.30	**KANAMA PANELİ-8: OBSTETRİK KANAMALAR / VAKA SUNUMLARI / SÖZEL BİLDİRİLER-16 ve SORU-CEVAPLAR**
	Panel Başkanları: Ali Acar, Mehmet Zeki Taner, Mete Tanır
	Panelistler: İsmail Özdemir, Ragıp Atakan Al, Berkan Sayal, Senem Yaman Tunç, Levent Keskin, Turhan Çağlar, Ayhan Sucak, Selçuk Özden, Fırat Öktem
	- Postpartum kanamada risk faktörleri, etyoloji, **Ayhan Sucak** - Sezaryen skar gebeliği. Tanı yönetim., **Turhan Çağlar** - Sezaryen skar gebeliği yakın izlem ile başarılı gebelik mümkün mü?, **Senem Yaman Tunç** - Doğum travmalarına bağlı kanama tanı ve yönetimi - olgu sunumu, **Hüseyin Levent Keskin** - Derin vajen yırtığı, paravaginal hematom olgularında VAC kullanımı, **Ragıp Atakan Al** - Kompresyon sütürleri ve atonide acar sütür tekniği, **Berkan Sayal** - Uterin devaskülarizasyon / cerrahi ve radyolojik, **Selçuk Özden** - Pas tanı ve yönetimi, sezaryen histerektomi, uterus rüptürü uterus koruyucu cerrahi, **İsmail Özdemir** - Cell salvage kullanımı, **Selçuk Özden** - Obstetrik kanamanın önlenmesi, kanamaya hazırlık: Gebelikte, doğum sırasında, doğumdan sonra, **Fırat Ökmen**
	SS-016 Sezaryen skar gebeliklerinin kısa dönem cerrahi tedavi sonuçları İrem Özaydın Kalaycı
17.30-19.00	**FETAL ANOMALİLER PANELİ-9: PERİNATAL ENFEKSİYONLAR, SENDROMLAR-SEKANSLAR ve PLASENTA-KORD-AMNİYOTİK SIVI SORUNLARI / VAKA SUNUMLARI / SÖZEL BİLDİRİLER 17 VE SORU-CEVAPLAR**
	Panel Başkanları: Saadettin Güngör, Metin İngeç, Mehmet Tayyar
	Panelistler: Arda Lembet, Başak Baksu, Başak Yıldırım, Mehmet Murat Işıkalan, Mehmet Güney, Oktay Kaymak, Pınar Tokdemir Çalış, Fatih Akkuş, Mucize Eriç Özdemir
	- CMV Yönetim Labirentinde Bir Vaka: Doğru Yolu Nasıl Bulduk?, **Fatih Akkuş** - Geç Gelen Tehlike: Doğuma Yakın Saptanan İki Toksoplazma Vakası'nın Öğrettikleri, **Mehmet Güney** - Sıradan Bir Virüs, Sıra Dışı Bir Vaka: Gebelikte İnfluenza Tanısının Perde Arkası, **Başak Yıldırım** - Özefagus Atrezisi ile Prezente Smith-Lemli-Opitz Sendromu, **Mucize Eriç Özdemir** - Non-immun hidrops fetalis: WES bulgularıyla İki Olgu, **Arda Lembet** - Anne Karnındaki Sessizlik: Polihidramniyosun Ardındaki Fetal Akinezi Gizemi, **Pınar Tokdemir Çalış** - Plasentadaki Saatli Bomba: Koranjioma Vakası'nın Kritik Yönetimi, **Başak Baksu** - ATAD3A Mutasyonu İle Tanımlanan Nadir Bir Fetal Akinezi Olgusu, **Mehmet Murat Işıkalan** - Görünmez Tehlike Vasa Previa: Tanı konulmasaydı Ne Olurdu?, **Oktay Kaymak**
	SS-017 Gebelikte sitomegalovirüs taraması: Üçüncü basamak bir merkezin iki yıllık deneyimi Nimet Alyörük Geçici
19.00-19.20	**Kapanış**

SALON B	1 KASIM 2025
10.30-12.00	**SÖZEL BİLDİRİLER OTURUMU - 3** **Oturum Başkanları:** Seval Erdinc, Semih Tugrul
10.30-12.00	**SS-049 Preeklampsi öngörüsünde plasental lösemi inhibitör faktörü (LIF) ve vasküler endotelyal büyüme faktörü (VEGF) düzeylerinin tanısal değeri ve birlikte kullanımının değerlendirilmesi** Ekin Asanoğlu Ekici **SS-050 Geç preterm bebeklerde antenatal kortikosteroidlerin fetal tiroid fonksiyonları üzerindeki etkilerinin retrospektif değerlendirilmesi** Yasin Semih Ekici **SS-051 Fetal tip 2 umbilikal-portal venöz sistemik şant: 7 vakanın prenatal yönetimi ve sonuçları** Dilek Buldum **SS-052 Birinci trimesterde kistik lezyon ile prezente olan fetüste intrahepatik venöz dönüş anormalliği: vaka sunumu** Görkem Arıca **SS-053 12. haftada artmış nukal saydamlık ile prezente olan fetal kardiyak miksoma: literatürde bildirilen en erken vaka** Elif Ayan Avcı **SS-054 Kardiyak tümör tanısı alan fetüslerin prenatal bulguları ve postnatal seyri** Hale Özer Çaltek **SS-055 Fetal aritmiler: supraventriküler taşikardi tanı ve yönetimi – iki vaka sunumu** Selçuk Atalay **SS-056 Cantrell pentalojisine eşlik eden amelia: vaka sunumu** Engin Öztürk **SS-057 Ebstein anomalisi prenatal ultrasonografi ile tanı: vaka sunumu** Begüm Aktaş Çiftçi **SS-058 PIEZO1 gen mutasyonu ile ilişkili hidrops fetalis vaka sunumu** Esin Şahin Toruk **SS-059 Multikistik ensefalomalasinin prenatal tanısı ve klinik yönetimi: vaka sunumu** Mehtap Korkut **SS-060 Prepontin araknoid kistlerin prenatal tanısı: iki vaka sunumu ve literatür incelemesi** Gunel Aliyeva **SS-061 Nöral tüp defektlerinin nadir bir sunumu: izole frontal ensefalosel prenatal tanısı** Esin Şahin Toruk **SS-062 Dandy–Walker malformasyonu prenatal tanısı ve perinatal sonuçları: üç yıllık tek merkez vaka serisi** Tamer Altındağ **SS-063 İniensefali: nadir vaka sunumu** Aziz Kından
13.30-15.00	**SÖZEL BİLDİRİLER OTURUMU - 4** **Oturum Başkanları:** Cihan İnan, Ebru Davutoğlu

13.30-15.00	**SS-064 Sessiz uterin skar dehisansının sonografik görünümü ve intraoperatif doğrulaması** Ayse Nur Işik Aydin **SS-065 Amniyotik bant sendromu: prenatal tanı zorluklarını vurgulayan vaka serisi** Sevil Cicek **SS-066 Dev fetal lenfanjiyom: vaka sunumu** Ozlem Aldemir Bukagikiran **SS-067 Artrogripozis multipleks konjenita: prenatal tanıdan genetik doğrulamaya nadir bir vaka** Yasemin Beyza Kaya Parspancı **SS-068 Prenatal olarak saptanan fetal adrenal hematom: vaka sunumu** Pelin Karalar Yıldız **SS-069 Binder sendromu benzeri yüz profili prenatal tespiti: vaka sunumu** Zühal Altıntaş **SS-070 İkiz gebelikte bilateral konjenital diz çıkığı: vaka sunumu** Esra Nuhoğlu Bilgin **SS-071 Şiddetli fetal anemi ve hidropsa yol açan sessiz plasental dekolman sonucu ani fetal kötüleşme** Öykü Yılmaz **SS-072 Nadir bir McKusick-Kaufman sendromu vaka sunumu** Gunel Aliyeva **SS-073 Protein tirozin kinaz 7 (PTK7) geninde homozigot mutasyon: vaka sunumu** Esin Şahin Toruk **SS-074 APO A5 homozigot mutasyonu ile ilişkili ciddi ailesel hipetrigliseridemi ve kronik pankreatitli gebelikte prenatal yönetim: vaka sunumu** Muradiye Yıldırım **SS-075 Pulmoner hemoraji ile komplike Takayasu arteriti olan gebelik: vaka sunumu** Esra Nuhoğlu Bilgin **SS-076 Gebelikte ve postpartum dönemde görülen Bell's paralizisi:Olgu serisi** Şule Birol İnce **SS-077 Ekstrapulmoner sekestrasyonun prenatal tanısı: iki vaka sunumu** Özge Öztürk **SS-078 Sağ aortik ark ve sağ duktus arteriyozus: vaka sunumu** Aybüke Fatma Dirgen Yakar

SOZLU BILDIRILER

SS-001

First-trimester ventriculothalamic angle as a novel predictor of fetal growth restriction: a prospective cohort study

Yasemin Dadaş[1], Zeynep Asli Oskovi Kaplan[1], Atakan Tanaçan[2], Fatma Doğa Öcal[2], A Seval Özgü Erdinç[2]
[1]University of Health Sciences, Ankara City Hospital Gynecology and Obstetrics Department
[2]University of Health Sciences, Ankara City Hospital Perinatology Department

OBJECTIVE: This prospective cohort study examined first-trimester ultrasound markers—nuchal translucency (NT), nasal bone (NB), and ventriculothalamic angle—to predict fetal growth restriction (FGR) or macrosomia later in pregnancy.

METHODS: A total of 511 healthy pregnant women underwent screening between 11+0 and 13+6 weeks gestation, with follow-up at 28-32 weeks. NT, NB, and ventriculothalamic angle were measured during the first trimester in fetal midsagittal sections by a single operator. The ventriculothalamic angle was defined as the angle between a line tracing the posterior surface of the brainstem (bordering the brainstem and fourth ventricle) and another line extending along the cranial border of the mesencephalon and thalamus (Figure 1). Fetal biometry and Doppler studies were performed at the follow-up visit.

RESULTS: Early-onset FGR was detected in 10 of the 511 women. These women formed the "patient" group, while the remaining participants were the "control" group. A ventriculothalamic angle of 91.5 degrees or above had 70% sensitivity and 98.6% specificity for predicting early-onset FGR.

There were significant differences in fetal biometric parameters (BPD, AC, FL, EFW) between the patient and control groups, with the control group having higher mean values. Patients with a ventriculothalamic angle ≥91.5 degrees were more likely to have associated fetal anomalies, including microcephaly, congenital heart defects, and hydrops fetalis.

CONCLUSION: Ventriculothalamic angle measurement at 11-14 weeks shows potential as an early predictor of FGR. Additional studies are necessary to confirm these findings and define clinical uses.

Keywords: ventriculothalamic angle, screening, fetal growth restriction

SS-002

Neurodevelopmental outcome in prenatally diagnosed isolated abnormalities of the corpus callosum

Cem Dagdelen[1], Tuğçe Tunç Acar[1], Aytekin Uzkar[1], Dilşen Kavuz[1], Cem Yaşar Sanhal[2], Mehmet Şimşek[1], İnanç Mendilcioğlu[1]
[1]Department of Gynecology Obstetrics, Division of Perinatology, Akdeniz University, Antalya, Turkey
[2]Private Perinatology Clinic, Antalya, Turkey

OBJECTIVE: Our aim was to investigate cases with isolated abnormalities of the corpus callosum(aCC) and report perinatal neurodevelopmental(ND) outcomes.

METHOD: In this single-center case series, we retrospectively analyzed the postnatal neurodevelopmental outcomes of patients followed up in our clinic for prenatal corpus callosum (CC) abnormalities between January 2016 and July 2025. Cases prenatally diagnosed with isolated CC anomalies were included in the study. When evaluating children born with aCC, fetuses with motor deficits that impair walking ability and severe behavioral disorders (autism spectrum disorder) were categorized as severe ND retardation, motor deficits that did not affect walking skills were categorized as moderate ND retardation, and clumsiness and mild speech disorders were categorized as mild ND retardation.

RESULTS: A total of 47 pregnancies with fetal CC malformations were identified. When the non-isolated, pregnancies terminated, and intrauterine ex fetuses were excluded, 12 fetuses were identified as live-born. The surviving children were enrolled in the ND follow-up program at our institution. Of the 12 children included in the analysis, three (25%) had mild ND impairment, five (41%) had normal ND outcomes, and four had severe ND retardation. Of the four children with severe ND impairment, two had complete CCA and two had partial CCA. Of the four children with severe neurodevelopmental impairment, two had complete CCA, and two had partial CCA.

CONCLUSION: While counseling parents in cases with corpus callosum anomalies during prenatal diagnosis, it should be taken into account that severe NE retardations may also occur in cases with partial CC anomalies.

Keywords: corpus callosum, Neurodevelopmental outcomes in children, prenatally diagnos

SS-003

How do we close the uterus in a cesarean section? An online survey trial

Gurcan Turkyilmaz
Lokman Hekim University, Faculty of Medicine, Department of Obstetrics and Gynecology

OBJECTIVES: The uterine closure technique in cesarean section (CS) has a significant impact on healing and niche formation; however, there is still no consensus on the optimal method. Due to a lack of robust recommendations, numerous techniques are frequently employed in practice. We aimed to investigate the frequency of techniques used and the factors affecting them.

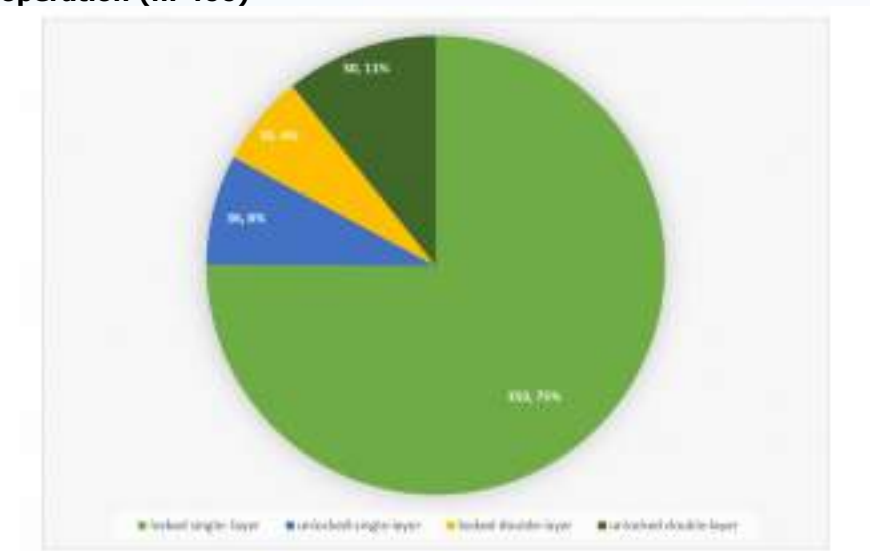
METHODS: An anonymous online survey in Turkish with 12 questions was sent to available obstetricians in Turkey. The primary response of interest was the type of uterine closure technique favored in CS. Secondary reactions of interest encompassed the formation of a bladder flap and closure of the peritoneum, rectus abdominis muscles, and subcutaneous tissues.

RESULTS: The primary approach utilized was the locked single layer at 75%, whilst the least preferred technique was the locked double layer at 6%. Also, 55.7% of physicians construct a bladder flap, whereas 80.8% routinely close the parietal peritoneum. 86% of surgeons typically choose to suture subcutaneous tissues when the thickness exceeds 2 cm. No significant difference was found in the frequency of uterine closure techniques between senior specialist physicians and less experienced obstetricians.

CONCLUSION: We showed that the predominant strategy for uterine closure in cesarean sections is the locked single-layer method. Moreover, most doctors seal the parietal peritoneum and stitch the subcutaneous tissue if it is thick. More efforts are necessary to ascertain the most effective uterine closure technique and to implement it consistently nationwide.

Keywords: cesarean section, niche, survey, technique, uterus

Graphic-1: Distribution of the procedure used for closing the uterus during a cesarean operation (n: 466)



Graphic-1: Distribution of the procedure used for closing the uterus during a cesarean operation (n: 466)

Table-1:Demographic features and characteristics of physicians (n: 466, (%))

Age (years)	25-35 35-45 45-55 55-65 >65	83 (17.8) 202 (43.3) 113 (24.2) 55 (11.8) 13 (2.7)
Years in practice	<5 5-10 10-15 15-20 >20	94 (20.1) 92 (19.7) 100 (21.4) 62 (13.3) 118 (25.3)
Title	Medical doctor Associate Professor Professor	360 (77.2) 50 (10.7) 56 (12.1)
Sex	Female Male	246 (52.7) 220 (47.3)
Affiliation	Public Hospital Education/Research Hospital University Hospital Private Hospital Private Clinic	70 (15) 113 (24.2) 84 (18.1) 115 (24.6) 84 ((18.1)
C/S operation number per year	<50 50-100 100-200 200-300 >300	115 (24.6) 110 (23.6) 110 (23.6) 56 (12) 75 (16.2)

Table-1:Demographic features and characteristics of physicians (n: 466, (%))

Table-2: Questions and answers regarding preferred techniques for Caesarean section in physicians (n:466 (%))

Question	Yes	No
Do you routinely create a bladder flap in a cesarean section?	206 (44.3)	260 (55.7)
Do you close visceral peritoneum routinely in cesarean section	92 (19.2)	374 (80.8)
Do you close parietal peritoneum routinely in cesarean section	298 (63.9)	168 (36.1)
Do you approximate rectus abdominis muscles routinely in cesarean section?	104 (22.3)	362 (77.6)
Do you suture subcutaneous tissues if more than >2 cm thickness routinely in cesarean section	401 (86)	65 (14)

Table-2: Questions and answers regarding preferred techniques for Caesarean section in physicians (n:466 (%))

Table-3: Distribution of uterine closure procedures stratified by physicians

Technique	≤10 years (n:186)	>10 years (n:280)	p value
Locked single layer	132 (71)	218 (77.8)	0.26
Unlocked single layer	16 (8.6)	20 (7.2)	0.19
Locked double layer	17 (9.1)	13 (4.6)	0.31
Unlocked double layer	21 (11.3)	29 (10.3)	0.21

Table-3: Distribution of uterine closure procedures stratified by physicians' years of expertise.

SS-004

Prenatal and Postnatal Follow-Up of Pregnancies with Fetal Intra-Abdominal Calcifications: A Single-Center Experience

Elif Ayan Avci, Büşra Tsakir, Gul Alkan bulbul, Hasan Berkan Sayal, And Yavuz
Department of Obstetrics and Gynecology, Antalya Training and Research Hospital

Fetal intra-abdominal calcifications (IAC) are rare findings on prenatal ultrasonography and represent a condition with a broad etiological spectrum. Isolated intrahepatic calcifications are usually benign and are most commonly attributed to vascular events or small parenchymal infarcts. In contrast, peritoneal or multi-organ calcifications may be associated with intrauterine infections, chromosomal abnormalities, gastrointestinal perforations, or tumoral lesions (1). The reported incidence is approximately 5–10 per 10,000 pregnancies (2).

In this study, 12 pregnancies with fetal IAC detected on prenatal ultrasonography over the past year were retrospectively evaluated. Maternal comorbidities included hypothyroidism (n=1), gestational diabetes mellitus (n=1), intrahepatic cholestasis (n=1), and gestational hypertension (n=1). TORCH serologies were negative in all cases; one patient had intermediate CMV IgG avidity, and one tested positive for Parvovirus IgM.

The localization of calcifications was as follows: isolated liver (n=5), spleen+liver (n=2), bowel+liver (n=2), stomach+diaphragm (n=1), liver+stomach (n=1), and intracranial+diaphragm+spleen (n=1). Four patients (33%) underwent invasive genetic testing and PCR analysis, while one underwent non-invasive prenatal testing; all results were normal.

Postnatal follow-up revealed one case with hepatic granuloma and one with growth restriction, while the remaining newborns were healthy. Our findings are consistent with previously published case series reporting favorable neonatal outcomes in isolated IAC (3,4). Some calcifications may become more evident postnatally, highlighting the importance of neonatal ultrasonographic follow-up (5).

In conclusion, isolated intrahepatic calcifications are the most frequent pattern and usually benign. In cases with multi-organ involvement, etiological investigations should be expanded, prenatal and postnatal assessments should follow a multidisciplinary approach.

Keywords: intra-abdominal calcifications, postnatal outcome, ultrasonography

figure 1

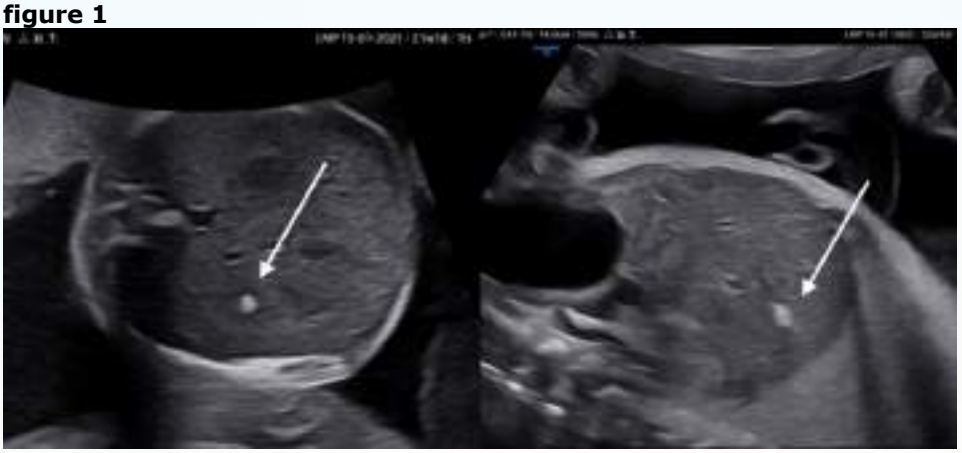


Figure 1. Ultrasound images of isolated fetal liver calcification (arrow) at 21 and 32 weeks of gestation.

figure 2

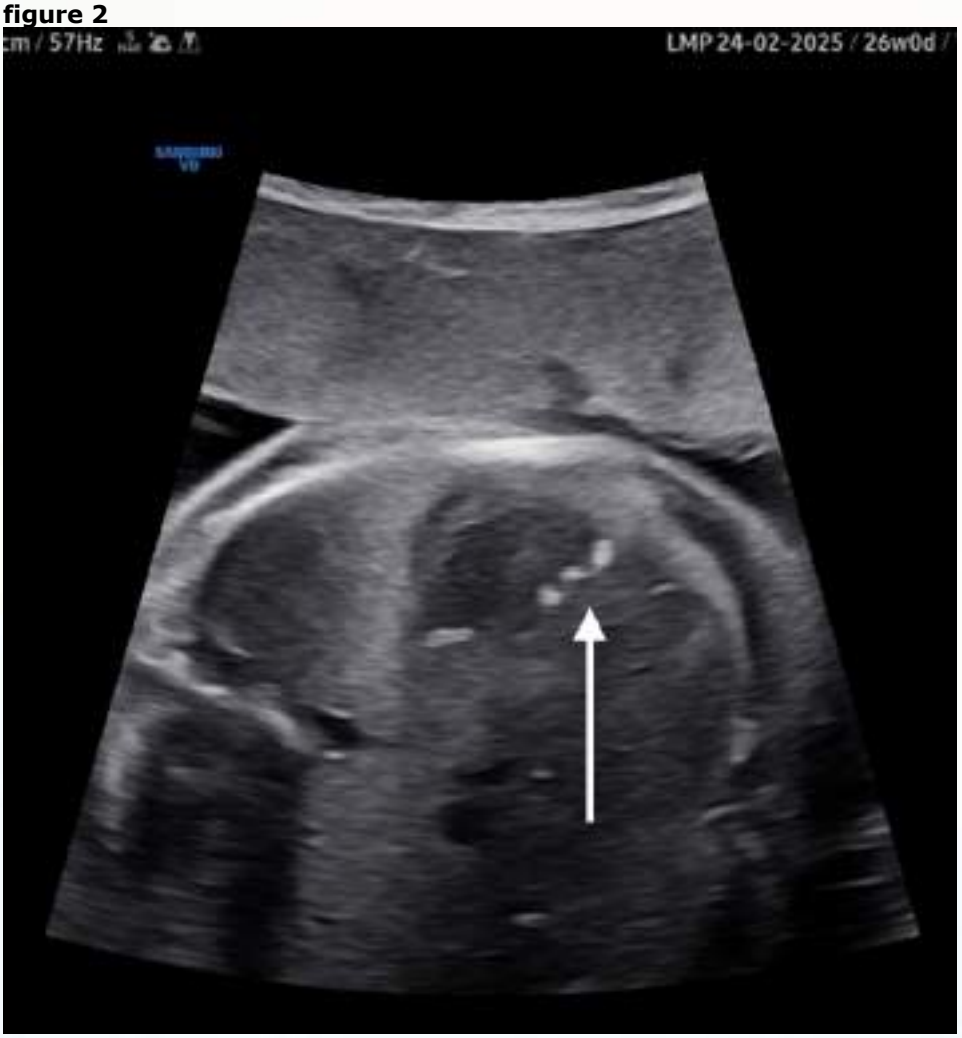


Figure 2. Ultrasound image showing multiple calcifications anterior to the fetal stomach (arrow) at 26 weeks of gestation.

SS-005

Fetal Yapısal Kardiyak Anomaliler Dışındaki Doğumsal Kardiyak ve Orta Mediasten Anomalilerin Perinatal Sonuçları

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AMAÇ: Prenatal dönemde fetal yapısal kardiyak anomaliler haricinde orta mediastende görülen kardiyak patolojilerin perinatal sonuçların değerlendirilmesi amaçlandı.

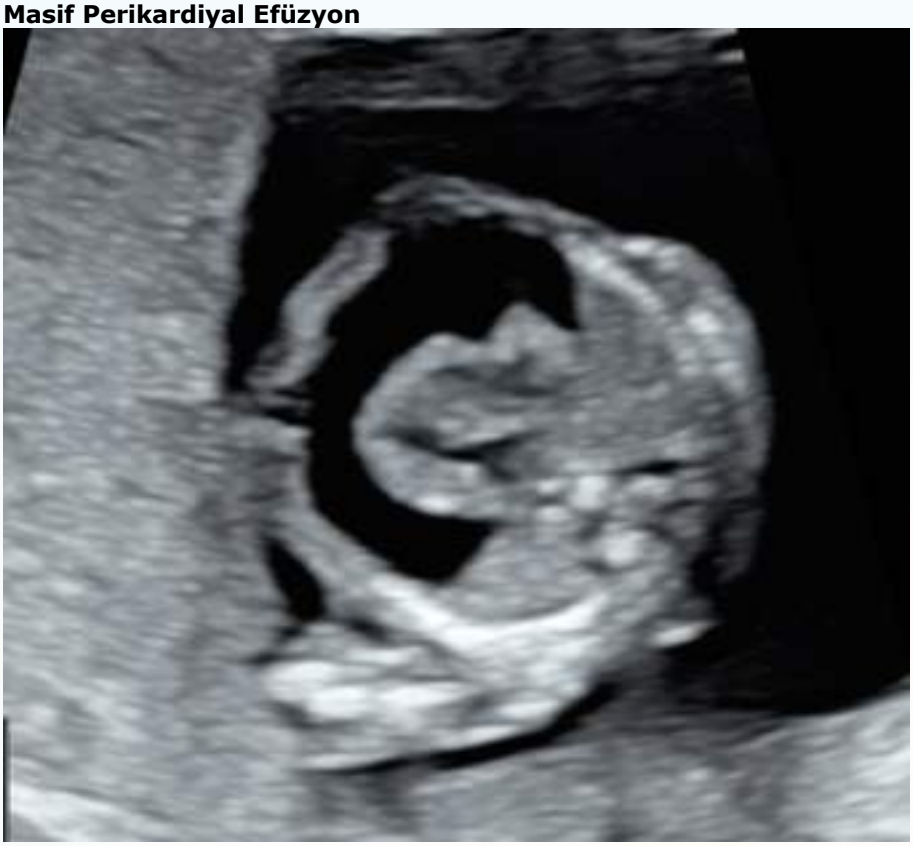
GEREÇ-YÖNTEM: 2021 Aralık-2025 Haziran tarihleri arasında perinatoloji kliniğine başvuran 62 olgu retrospektif değerlendirildi. 11 hasta monokoryonik ikiz gebelik olmasından dolayı çalışmadan çıkartıldı. Ultrasonografi muayenesinde tanı konulan olgulara prenatal genetik incelemede sitogenetik, array ve tüm ekzom analiz yapıldı. Postnatal tüm olgulara Pediatrik Kardiyoloji konsültasyonu planlandı.

BULGULAR: Değerlendirilen 51 olguya ortalama 21. gebelik haftasında tanı koyuldu. Olguların %94'ünde perikardiyal efüzyon (n=48), %8'inde kardiyak divertikül (n=4) ve %4'ünde kardiyak lenfatik malformasyon (n=2) saptandı. Tüm olguların %31'i izole perikardiyal efüzyon tanısı aldı (n=16). Diğer majör sistem anomalileri ile birliktelik %61 oranında bulundu (n=31). Bu olgulara en sık kardiyovasküler anomaliler eşlik etti (n=18). Olguların %27'sinde amniyon sıvısından parvovirüs ve sitomegalovirüs enfeksiyonu bakıldı (n=14). Dört olguda parvovirüs saptandı. %75 hasta prenatal genetik analiz yaptırmış olup (n=38); bu hastaların 8'inde genetik anomali saptadı. En sık saptanan genetik anomali Trizomi 21 olarak bulundu (n=2). İzole perikardiyal efüzyon bulunan olguların sadece birinde 10q21.3q22.'de delesyon saptandı. Prenatal dönemde divertikül tanısı konulan olguların %50'sinde genetik anormallik bulundu (n=2). Divertikül olan bir olguda 16p.13.11 mikrodelesyonu tespit edilmiş olup; diğer olguya ise Noonan sendromu tanısı konulmuştur. Olguların %37'si terminasyon (n=19), %39'u canlı doğum (n=20) ve %16'sı ölü doğum (n=8) ile sonuçlandı. Dört olgunun gebelik takibine devam edilmektedir.

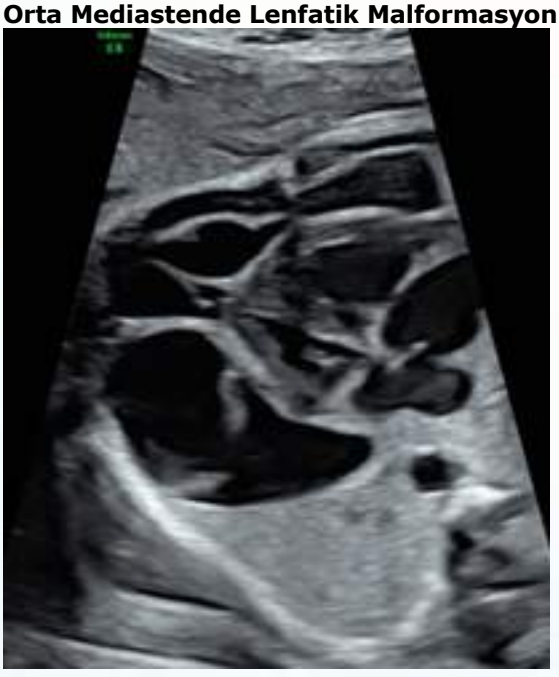
SONUÇ: İzole perikardiyal efüzyonlar perinatal dönemde genellikle daha iyi prognozlu seyrederken, ek majör yapısal anomali eşlik eden durumlarda prognoz farklılık gösterebilmektedir. Eşlik eden kromozom anomalileri açısından prenatal genetik analiz tüm olgulara önerilmelidir.

Anahtar Kelimeler: Perikardiyal efüzyon, divertikül, lenfatik malformasyon

Masif Perikardiyal Efüzyon



Orta Mediastende Lenfatik Malformasyon



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SS-006

Prediction of Delivery Time Using Intrapartum Ultrasound at the onset of Active Labor in Nulliparous Women

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OBJECTIVE: To investigate the predictive value of intrapartum ultrasound parameters—angle of progression (AoP), head–perineum distance (HPD), occiput position, head direction, and midline angle (MLA)—measured at the onset of the active phase of labor in nulliparous women.

METHODS: In this prospective study, 57 nulliparous women admitted to Akdeniz University Hospital between March 2024 and August 2025 were evaluated at 6 cm cervical dilatation. Labor duration, delivery outcomes, and neonatal data were recorded. Statistical analyses included Spearman correlation, multivariable regression, and ROC curve analysis.

RESULTS: AoP showed a significant negative correlation with labor duration ($r = -0.394$; $p = 0.002$). For predicting delivery within 2 hours, AoP had an AUC of 0.692 (95% CI: 0.548–0.836; $p = 0.009$), with 62.5% sensitivity at a cut-off ≥124.5°. For predicting delivery within 4 hours, AoP yielded an AUC of 0.761 (95% CI: 0.616–0.905; $p < 0.001$) at a cut-off ≥119.5°, with 65% sensitivity and 93.8% PPV. HPD correlated positively with labor duration and excluded early delivery at ≤39.5 mm (negative likelihood ratio = 0). Head station was the strongest independent predictor ($\beta = 0.258$; $p = 0.017$), with significantly higher delivery rates within 2 hours when station was ≥+1 ($p < 0.001$). Occiput anterior and upward head direction were associated with shorter labor, whereas prolonged labor correlated with lower Apgar scores. MLA showed no significant predictive value.

CONCLUSION: Intrapartum ultrasound at the onset of active labor provides clinically valuable parameters—particularly AoP, HPD, and head station—for predicting labor duration in nulliparous women.

Keywords: intrapartum ultrasound, prediction of delivery time, nulliparity

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SS-007

Maternal and fetal outcomes of breast stimulation for augmentation/induction of labor in term pregnancies

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INTRODUCTION AND AIM: Aim of this study is to show that nipple stimulation for labor induction/augmentation in term pregnancies has an active contribution to labor and significantly reduces the level of pain during labor.

MATERIALS-METHODS: This prospective study was conducted between 01/07/2023 and 01/02/2024 in the labor and delivery room of T.C. Sakarya University Research and Training Hospital. It was conducted with 110 patients with cervical dilatation between 3 and 4 cm, Bishop score <6, and insufficient uterine activity (<3 contractions in 10 minutes), who were admitted to the delivery for the purpose of initiating labor or labor. The contribution of nipple stimulation to labor was evaluated in patients. Membrane stripping was applied to 56 cases in the control group while 3 cycles of breast stimulation were applied to 54 cases in the study group in addition to membrane stripping.

RESULTS: At the end of the first and second cycle in the breast stimulation group, Bishop score was significantly higher than the control group (p=0.018 and p=0.031). The need for induction is less in the stimulation group than in the control group (p<0.001), and the VAS score also decreased significantly in the breast stimulation group (p=0.03).

CONCLUSION: Nipple stimulation has been observed to improve Bishop score, reduce the need for synthetic induction, reduce pain during active labor, and accelerate lactation. Nipple stimulation should be administered under supervision as it may cause fetal distress. Studies with larger case series should be conducted to standardize nipple stimulation application.

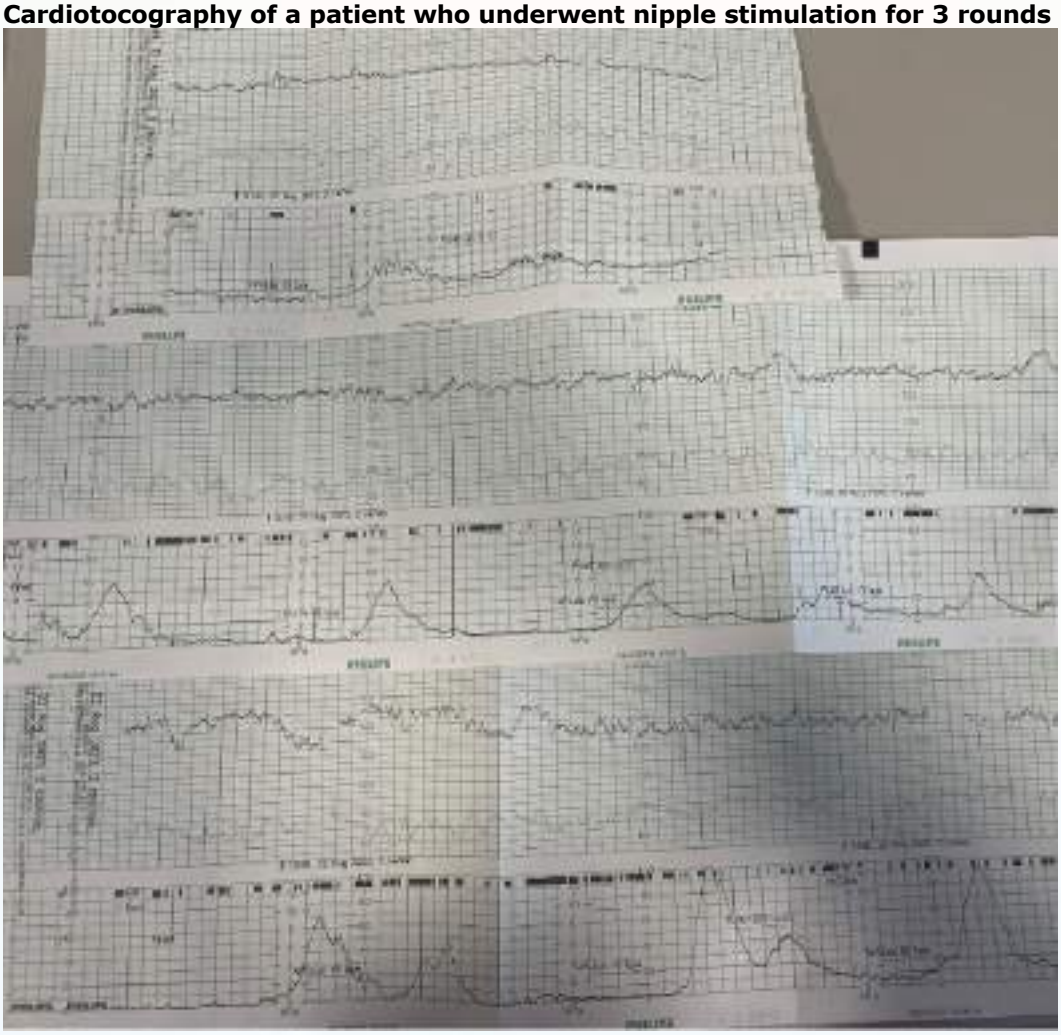
Keywords: Induction,labor,nipple stimulation

Bishop score changes between control and nipple stimulation group

Bishop Skoru	Kontrol (n=56)	Meme Stimülasyonu (n=54)	[2]p-değeri
Başlangıç	5 [4-6] [a]	5 [5-6] [a]	0,264
İlk Kürün Sonunda	6 [5-6] [a,b]	6 [5,75-7] [a,b]	0,264
İkinci Kürün Sonunda	6 [5-6] [b]	6 [6-7] [a,b]	0,264
Üçüncü Kürün Sonunda	6 [5-6] [b]	6 [5,75-7] [b]	0,264
[1]p-değeri	<0,001 *	<0,001 *	0,264

İstatistikler aritmetik ortanca [çeyreklikler arası genişlik] biçiminde gösterilmiştir.
1: Başlangıç ve kür zamanları arasındaki karşılaştırmalara ilişkin p-değerleri, 2: Başlangıç ve kür zamanları arasındaki değişim yönünden kontrol ve meme stimülasyonu grupları arasındaki karşılaştırmalara ilişkin p-değeri
**: Aynı üst indise (harf) sahip periyotlar arasında fark yoktur, farklı üst indise sahip periyotlar arasında ise istatistiksel olarak anlamlı fark vardır.*

Cardiotocography of a patient who underwent nipple stimulation for 3 rounds



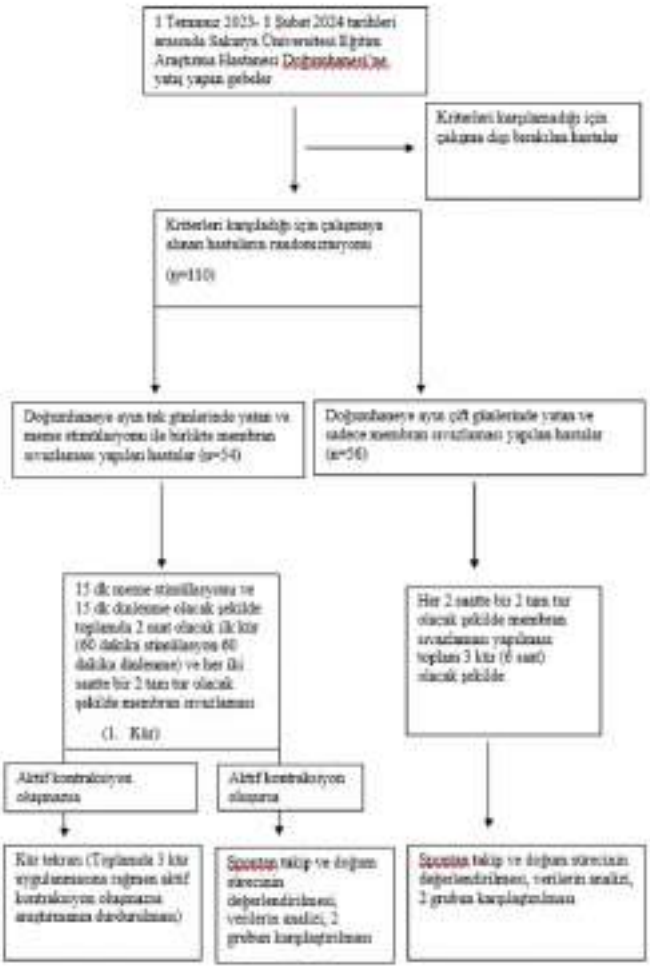
Clinical characteristics of the control and nipple stimulation groups

Tablo 4.2. Kontrol ve meme stimülasyonu gruplarının klinik özellikleri.

			Kontrol (n=56)		Meme Stimülasyonu (n=54)	p-değeri
Bishop Skoru	Başlangıç	56	5 [4-6]	54	5 [5-6]	**0,048**
	İlk Kürün Sonunda	56	6 [5-6]	54	6 [5,75-7]	**0,018**
	İkinci Kürün Sonunda	50	6 [5-6]	41	6 [6-7]	**0,031**
	Üçüncü Kürün Sonunda	44	6 [5-6]	26	6 [5,75-7]	0,168
Yeterli Kontraksiyona Ulaşma Zamanı (dk)		14	180 [105-240]	33	120 [120-240]	0,830
Yeterli Kontraksiyona Ulaşılan Kür Sayısı		14	2 [1-2]	33	2 [1-2]	0,960
Ulaşıldığı Andaki VAS Skoru		13	9 [8,5-10]	33	8 [7-9]	**0,003**
Doğumun İlk Evresinin Süresi		13	180 [120-375]			
Doğumun İkinci Evresinin Süresi		13	15 [11,5-45]	29	15 [10-30]	0,610
Doğumun Üçüncü Evresinin Süresi		13	15 [15-15]	29	15 [15-15]	0,501
APGAR 1. Dakika		13	9 [9-9]	29	9 [9-9]	1,000
APGAR 5. Dakika		13	10 [10-10]	29	10 [10-10]	1,000
Laktasyon Tatmin Skoru		13	9 [8,5-9]	29	9 [9-9,5]	0,185
Laktasyon Başlangıcı (Dakika)		13	15 [15-22,5]	29	0 [0-15]	**0,001**

İstatistikler aritmetik ortalama ±standart sapma ve ortanca [çeyreklikler arası genişlik] biçiminde gösterilmiştir.

Flow chart



Reasons of cesarean delivery in induction and non induction group

Tablo 4.5. Sezaryen ile doğum yapan hastalarda indüksiyon verilen ve verilmeyen gruplara göre sezaryen doğum nedenlerinin dağılımı.

Grup	İndüksiyon Verildi mi?	CS doğum nedenleri		p-değeri
		İlerlemeyen Travay	Fetal Distress	
Kontrol	Hayır	0 (0)	0 (0)	-
	Evet	6 (75)	2 (25)	
Meme Stimülasyonu	Hayır	2 (66,7)	1 (33,3)	0,429
	Evet	4 (100)	0 (0)	

İstatistikler sayı (%) biçiminde gösterilmiştir.

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SS-008

MATERNAL and NEONATAL OUTCOMES of SUBSTANCE USE in PREGNANCY: A RETROSPECTİVE COHORT

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AİM: Substance abuse is a major public health issue. In Turkey, the prevalence of substance use has been reported as 6.4%. Opioid use during pregnancy is associated with increased maternal and perinatal morbidity and mortality, most notably preterm birth, with a relative risk of 3.45. This study evaluated the demographic features, obstetric complications, and neonatal outcomes of pregnant women with substance use disorder.

MATERİALS AND METHODS: A retrospective study was conducted on 18 substance-using mothers and their neonates delivered at Ankara Etlik City Hospital (2023-2024). Data on demographics, substance use patterns, comorbidities, and neonatal abstinence syndrome (NAS) were analyzed using the Finnegan scoring system.

RESULTS: Mean maternal age was 28.6 ± 6.45 years; 94.4% smoked tobacco. Mean substance dependence duration was 7.7 ± 4.0 years. Poly-substance use was predominant (55.6%), followed by opioids (27.8%) and amphetamines (16.7%). Most (61.1%) used substances throughout pregnancy. Infectious comorbidities included hepatitis C (16.5%), syphilis (11%), and HPV (5.5%). Preterm labor (66.7%) and oligohydramnios (33.3%) were the main obstetric complications. Mean gestational age was 32.6 ± 5.3 weeks; mean birth weight was 2166 ± 889 g. Three neonates with Finnegan scores ≥12 or seizures received phenobarbital for NAS. Four infants remained with their mothers under supervision; others entered child protection. One-third of mothers had psychiatric comorbidities.

CONCLUSİON: Substance use in pregnancy is strongly linked to preterm birth, NAS, and infectious risks. Poly-substance use and psychiatric issues worsen outcomes. Early detection, multidisciplinary care, and systematic neonatal screening are vital for improving health in this high-risk group.

Keywords: Neonatal Abstinence Syndrome, Pregnancy Outcome, Substance Use Disorder

Maternal, Obstetric, and Clinical Characteristics Associated with Substance Use During Pregnancy

N=18	N %	MEAN ± SD	MEDİAN (25. and 75. percentil)
Maternal Age		28.6 ± 6.45	27.0 (24.3, 32.5)
Body Mass Index		27.9 ± 5.5	29.0 (25.0, 32.0)
Smoking Status	17 (94.4%)		
Marital Status	6 (33.3%)		

Comorbidities			
HCV positivity	3 (16.5)		
HPV positivity	1 (5.5)		
Syphilis	2 (11.0)		
Psychiatric comorbidity	3 (16.6)		
Obstetric History			
Gravida		2.7 ± 1.7	3.0 (1.0, 3.0)
Parity		1.3 ± 1.4	1.0 (0.0, 2.0)
Abortus		1.2 ± 1.4	0.0 (0.0, 0.0)
Living children		0.1 ± 0.3	1.0 (0.0, 2.0)
D&C (Dilation and Curettage)		0.2 ± 0.5	0.0 (0.0, 0.0)
Previous delivery mode			
Number of cesarean sections		0.9 ± 1.5	0.0 (0.0, 1.0)
Vaginal delivery		0.3 ± 0.5	0.0 (0.0, 0.7)

Values are expressed as mean ± standard deviation (SD), median (interquartile range), or number (percentage). Abbreviations: SD, standard deviation; D/C, dilation and curettage; HCV, Hepatitis C virus; HPV, Human papillomavirus.

Substance Use During Pregnancy: Duration, Type, Trimester of Use, and Healthcare Utilization

N: 18	N (%)	NEAN ± SD	MEDİAN (25. Ve 75. percentil)
Duration of Dependence (years)		7.7 ± 4.0	7.5 (4.0, 10.8)
Type of Dependence			
Opioid	5 (27.8)		
Amphetamine	3 (16.7)		
Polysubstance	10 (55.6)		
Substance Use by Trimester			
Use throughout pregnancy	11 (61.1)		
Intermittent use	3 (16.7)		
Use during first two trimesters	4 (24.4)		
Number of outpatient clinic visits during pregnancy		1.7 ± 1.8	1.0 (0.0, 3.0)
Number of emergency department visits during pregnancy		3.2 ± 3.8	2.0 (1.0, 3.7)

Data are presented as mean ± standard deviation (SD), median (25th–75th percentile), or n (%). SD: Standard deviation

Obstetric and Neonatal Outcomes Associated with Substance Use During Pregnancy

N: 18	N (%)	MEAN ± SD	MEDİAN (25. and 75. percentil)
Oligohydramnios	6 (33.3)		
Placental abruption	1 (5.6)		
Preterm labor	12 (66.7)		
Postpartum hemorrhage	5 (27.8)		
Mode of delivery			
Cesarean section	11 (61.1)		
Vaginal delivery	7 (38.9)		
Postpartum maternal intensive care	2 (11.1)		
Gestational week at delivery		32.6 ± 5.3	34.0 (31.5, 36.5)
Birth weight		2166 ± 889	2305 (2120, 2679)
Apgar score at 1 minute		5.7 ± 2.9	7.0 (4.2, 7.7)
Apgar score at 5 minute		6.8 ± 3.3	8.0 (7.2, 8.7)
Neonatal Abstinence Syndrome	3 (16.6)		

Data are presented as mean ± standard deviation (SD), median (25th–75th percentile), or n (%). SD: Standard deviation

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SS-009

Two Different Approaches To Advanced-Stage Twin Anemia Polycythemia Sequence Cases

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INTRODUCTION: Treatment of twin anemia polycythemia sequence (TAPS) options include transfusion, selective reduction and fetoscopic laser fotocoagulation (FLF).In this report we present the cases of two different methods.

Case 1: Our case with the first treatment method that we used: Age 26, gravid 2, MCDA twin was referred our clinic in 19th week with pre-diagnosis selective growth restriction. At 22 weeks, the donor's middle cerebral artery (MCA) peak systolic velocity (PSV) was 2.25 MoM, while the recipient's was 0.67 MoM additionally pericardial effusion was detected Selective reduction planned for the patient evaluated stage 3 TAPS. Selective fetal reduction was performed in the recipient using intrafetal laser. Subsequent follow-ups showed a decrease in MCA-PSV in the donor.Currently, the fetus is in its 32nd week of pregnancy and pregnancy follow-up continues in our clinic.

Case 2: Age 34, gravid 2, twin was referred our clinic in 20th week with pre-diagnosis stage 3 TAPS. The recipient presented with hydrops and reversed a-wave in the ductus venosus. MCA-PSV were 0.63 MoM for the recipient and 1.85 MoM for the donor. FLF planned for the patient evaluated stage 3 TAPS.The recipient's amniotic fluid was entered, and after the anastomoses were coagulated, solominization was performed.Follow-up showed an MCA-PSV of 0.77 MoM, resolved cardiac decompremised in the recipient and the donor's MCA-PSV 1.4 MoM.The patient is currently at 23 weeks of gestation and remains under follow-up at our clinic.

CONCLUSION: In advanced-stage TAPS cases, the treatment option should be selected according to the patient's clinical status.

Keywords: Fetal anemia, laser photocoagulation, twin anemia polycythemia sequence

SS-010

Congenital Scimitar Syndrome: Prenatal Diagnosis and Case Report

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Sağlık Bilimleri Üniversitesi, Zeynep Kamil EAH, Perinatoloji Kliniği, İstanbul

INTRODUCTION: Scimitar syndrome is a rare congenital anomaly characterized by partial anomalous pulmonary venous return, most often with right pulmonary veins draining into the inferior vena cava. It may be associated with right lung hypoplasia or systemic arterial supply. Clinical presentation ranges from severe neonatal disease to incidental findings in adulthood. Prenatal diagnosis is uncommon but increasingly feasible with modern fetal echocardiography.

Case

A 24-year-old primigravida was referred at 28 weeks of gestation for mild midline displacement of the fetal heart. At 35 weeks, detailed echocardiography showed normal left pulmonary venous drainage into the left atrium, while the right pulmonary veins drained anomalously into the inferior vena cava via a scimitar vein. No systemic arterial supply or additional malformations were identified, consistent with isolated scimitar syndrome.

The infant was delivered at term without respiratory distress. Postnatal echocardiography confirmed anomalous right pulmonary venous drainage and normal left-sided return. The neonate remained asymptomatic, and conservative follow-up was planned.

Discussion

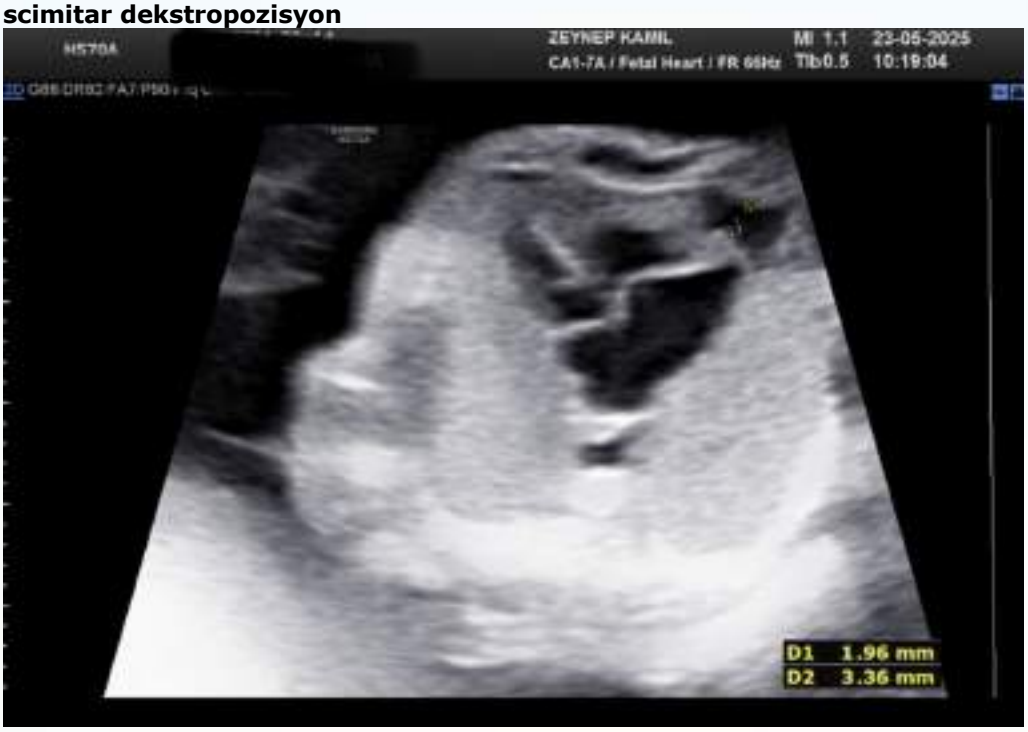
Scimitar syndrome has two clinical forms: an infantile type with respiratory distress, cyanosis, infections, or heart failure, and an adult form that is usually asymptomatic. Prognosis depends on associated anomalies; isolated cases typically have favorable outcomes.

Prenatal diagnosis requires systematic assessment of cardiac position and pulmonary venous drainage. In this case, despite the absence of dextroposition or pulmonary hypoplasia, careful fetal evaluation enabled early recognition. Prenatal detection guided postnatal confirmation, reassured the family, and supported tailored management.

CONCLUSION: Isolated scimitar syndrome can be diagnosed prenatally. Early recognition facilitates neonatal care and long-term follow-up.

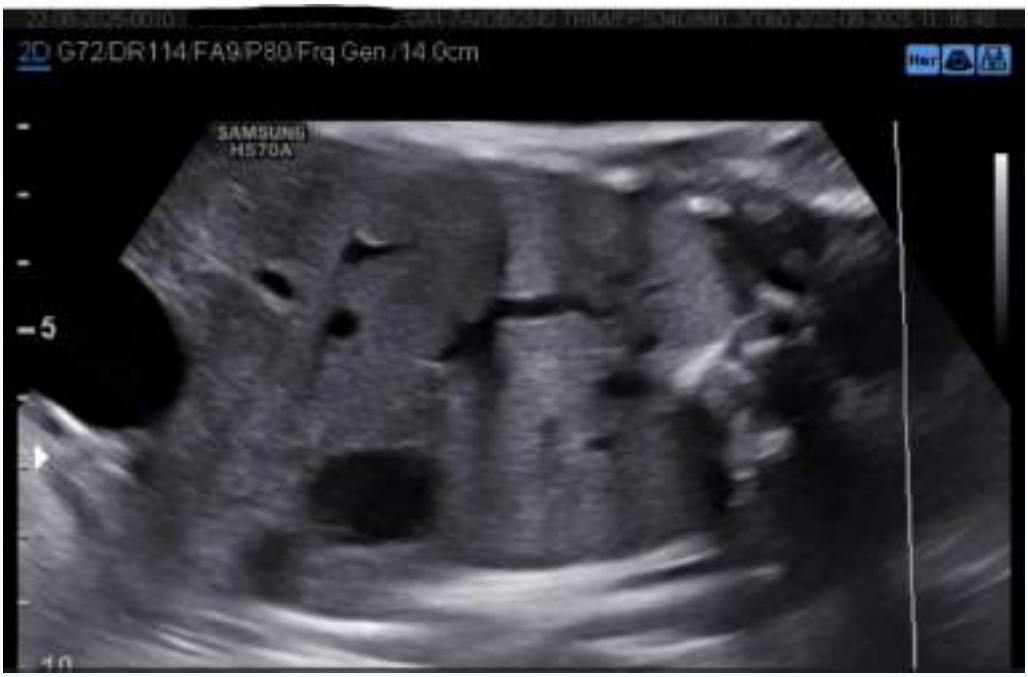
Anahtar Kelimeler: scimitar, papvc, dekstropozisyon

scimitar dekstropozisyon



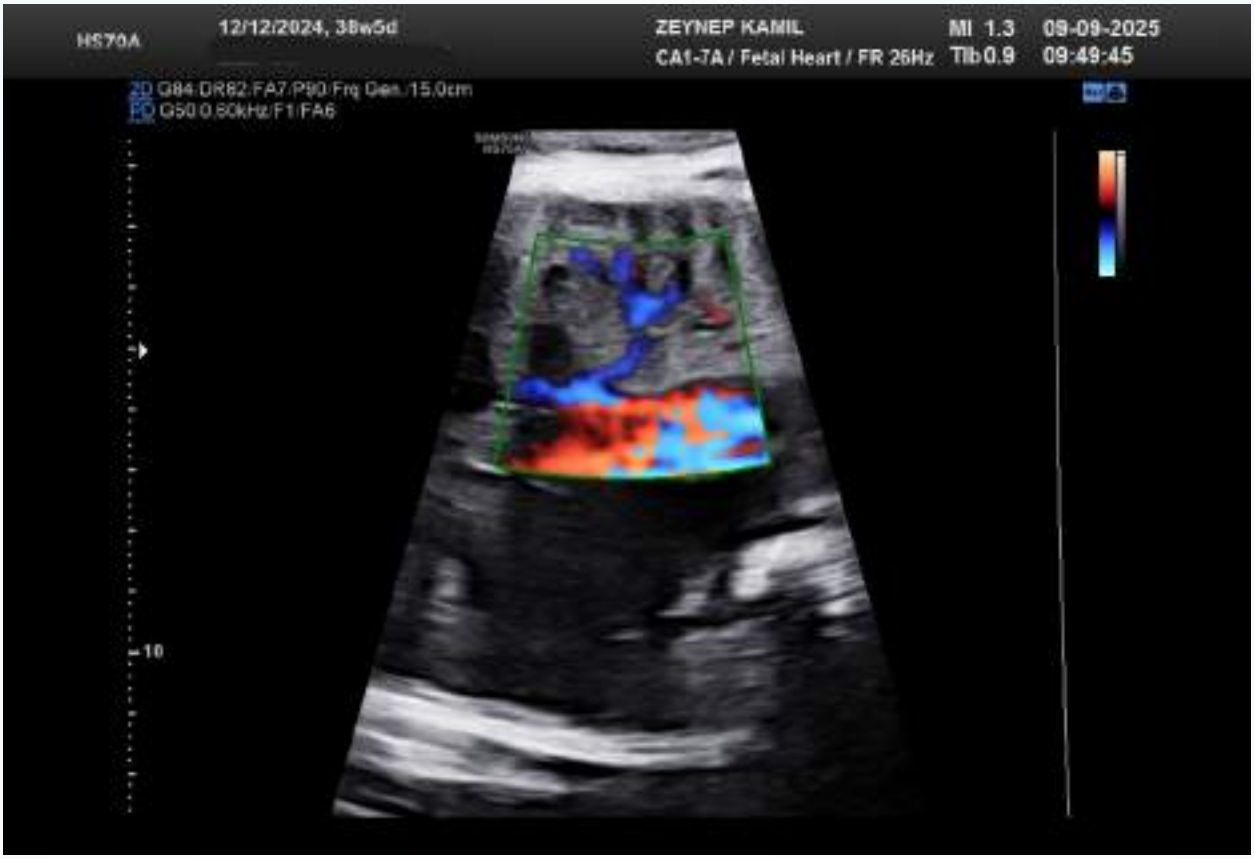
23. haftada dekstropozisyon ve perikardial efüzyon

scimitar veni



gri skalada scimitar veni

scimitar veni



sağ pulmoner venlerin birleşip scimitar veni olarak inferior vena cavaya drenajı

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SS-011

Recurrent molar pregnancy in a homozygous KHDC3L variant carrier

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INTRODUCTION: Recurrent molar pregnancy (RHM) is often associated with maternal-effect genes, notably NLRP7 and KHDC3L. We report a gravida 7 woman with multiple molar gestations and a rare homozygous KHDC3L variant.

Case Presentation: A 27-year-old (G7P1A5) presented at 15+ weeks' gestation with ultrasound revealing an anterior placenta with "snow-storm" appearance, suggestive of partial mole. Her history included five molar pregnancies and one intrauterine fetal demise at six months. Genetic testing identified a homozygous KHDC3L c.40A>G (p.Met14Val) variant of uncertain significance (VUS); paternal testing was negative. Previous IVF with pre-implantation genetic testing (PGT) excluded embryo homozygosity for this variant, yet recurrence occurred. The family declined invasive prenatal diagnosis and chose to continue the pregnancy.

DISCUSSION: KHDC3L encodes a subcortical maternal complex component; pathogenic variants can cause RHM via imprinting defects in oocytes. This variant is rare, absent in healthy homozygotes, and found in a patient with a highly suggestive phenotype, supporting possible pathogenicity. Literature indicates recurrence risk persists despite ART or PGT, and fetal heterozygosity does not cause disease. In our patient, maternal homozygosity likely predisposes to recurrent molar gestations. Counselling emphasised that future pregnancies remain high-risk and that PGT may not prevent recurrence. Screening of female relatives was recommended.

CONCLUSION: Maternal-effect gene analysis is valuable in RHM. This case underlines that KHDC3L homozygosity can be clinically relevant and that recurrence may occur despite PGT.

Keywords: KHDC3L, Maternal-effect gene, Recurrent molar pregnancy

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SS-012

Child outcomes following fetal intracranial hemorrhage

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INTRODUCTION: Fetal intracranial hemorrhage(FIH) is a rare but potentially serious event with an estimated incidence of 0.5 to 1 in 1000 pregnancies. The most common subtype of FIH is intraventricular hemorrhage (IVH) and can be graded from 1 to 4 based on the ultrasound grading of IVH in the fetus.

METHOD: We retrospectively analyzed the neurodevelopmental status of live-born fetuses in cases of fetal IVH diagnosed by fetal MRI and ultrasound at our clinic between 2016 and 2025.When evaluating children born with IVH, fetuses with motor deficits that impair walking ability and severe behavioral disorders (autism spectrum disorder) were categorized as severe ND retardation, motor deficits that did not affect walking skills were categorized as moderate ND retardation, and clumsiness and mild speech disorders were categorized as mild ND retardation

RESULTS: In a retrospective review, pregnancy termination was performed in 4 of 18 patients diagnosed with IVH, while information could not be obtained in 8 patients because they underwent neurological follow-up outside our hospital. ND follow-up of 6 patients aged between 1 and 8 years who were followed for IVH in our hospital was performed by pediatric neurologists.ND outcomes were normal in 2 of the 6 cases, severe neurodevelopmental retardation was observed in 3, and moderate neurodevelopmental delay was observed in 1. Of the 3 patients with severe neurodevelopmental deficits, 2 had grade 4 IVH, while 1 had grade 3 IVH.

CONCLUSION: This case series demonstrates that fetuses diagnosed with prenatal IVH are at high risk for adverse perinatal neurodevelopmental outcomes.

Keywords: fetal intracranial hemorrhage, neurodevelopmental outcome, Prenatal counseling parents

SS-013

Poliarteritis Nodosa Ve Preeklampsi Zemininde Gelişen Renal Hematom:Vaka Sunumu

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INTRODUCTION: Polyarteritis nodosa (PAN) is a necrotizing vasculitis affecting small- and medium-sized arteries. Although rare in pregnancy, it may cause severe maternal and fetal complications.

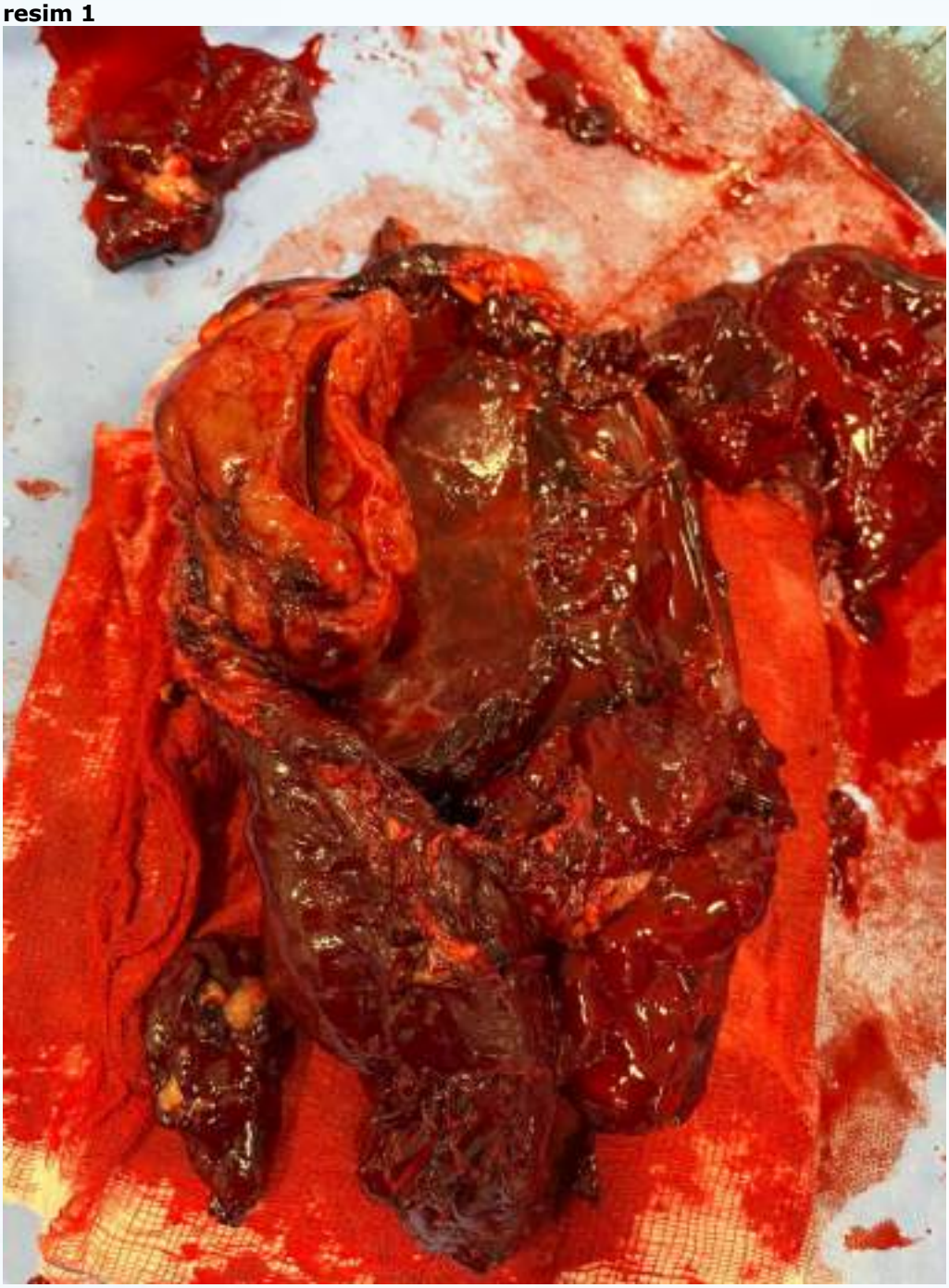
Case Presentation: We report a 33-year-old primigravida at 26 weeks of gestation admitted with elevated liver function tests and a history of hypertension from early pregnancy. During follow-up, she developed acute severe left flank pain at 28 weeks. Imaging revealed a large subcapsular renal hematoma. Due to hypovolemic shock and fetal bradycardia, emergency cesarean delivery was performed, resulting in the birth of a preterm infant with intraoperative demise. A retroperitoneal hematoma originating from the left kidney was identified, requiring nephrectomy. Histopathological evaluation revealed medium- and small-vessel vasculitis consistent with PAN. The patient recovered postoperatively and was referred to rheumatology for further management.

DISCUSSION: PAN in pregnancy is extremely rare, with fewer than 20 cases reported to date. Renal hematoma as a complication has been described in non-pregnant patients but, to our knowledge, has not previously been reported during pregnancy. PAN in pregnancy carries a high risk of maternal morbidity and mortality, and differentiation from hypertensive disorders of pregnancy remains challenging.

CONCLUSION: This is the first reported case of PAN in pregnancy complicated by renal hematoma. Clinicians should consider vasculitis in the differential diagnosis of atypical hypertensive disorders during pregnancy. Further studies are needed to better define management strategies and outcomes. Keywords: Polyarteritis nodosa, renal hematoma, preeklampsia

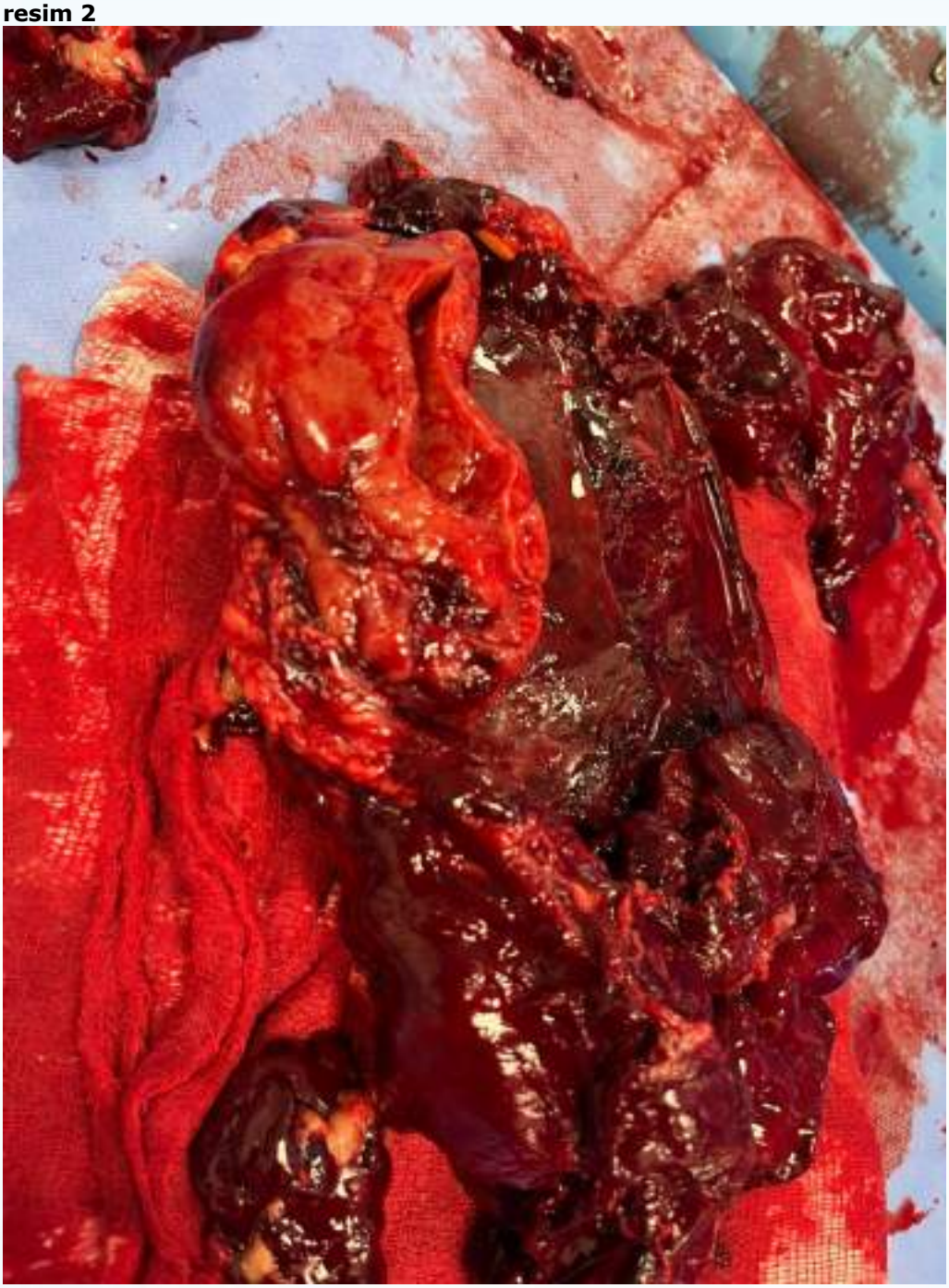
Anahtar Kelimeler: Polyarteritis nodosa, preeklampsia, renal hematoma

resim 1



renal parankim ve hematom görünümü

resim 2



renal parankim ve hematom görünümü

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SS-014

Follow-up and Perinatal Outcomes in Pregnancies with Discordant Abnormal NIPT and/or CVS Results and Normal First-Trimester Ultrasound Findings: A Case Series

EKREM ERGENÇ
Ekrem Ergenç

OBJECTIVE: To evaluate the follow-up and perinatal outcomes of pregnancies with discordant abnormal non-invasive prenatal testing (NIPT) and/or chorionic villus sampling (CVS) results despite normal first-trimester ultrasound (USG) findings.

METHODS: This retrospective case series included seven pregnant women referred to the Perinatology Clinic of Karadeniz Technical University between 2022 and 2024. All patients underwent NIPT and/or CVS, with subsequent amniocentesis performed in selected cases. Pregnancies showing discordance between abnormal NIPT/CVS results and normal first-trimester USG findings were analyzed for perinatal outcomes, including intrauterine growth restriction (IUGR), placental insufficiency, preeclampsia, fetal distress, gestational age at delivery, neonatal birthweight, and short-term neonatal results.

RESULTS: Among the seven cases, four developed IUGR, one had preeclampsia, and one showed placental insufficiency with fetal distress, while three pregnancies resulted in healthy neonates without complications. Despite abnormal NIPT and/or CVS results, amniocentesis findings were normal in most cases. Nevertheless, pregnancies remained at risk for adverse perinatal outcomes such as IUGR and hypertensive disorders. These findings suggest the role of confined placental mosaicism or other biological factors in generating discordant results between genetic testing and ultrasound.

CONCLUSION: Pregnancies with abnormal NIPT and/or CVS results but normal first-trimester USG findings require close follow-up, even if amniocentesis confirms a normal karyotype. Clinicians should remain cautious regarding possible complications including IUGR, preeclampsia, and placental insufficiency. Integration of ultrasound findings with molecular genetic testing may provide a more reliable strategy for prenatal management and counseling.

Keywords: NIPT, CVS, Ultrasound, Perinatal outcomes.

Clinical characteristics, genetic results, and perinatal outcomes of the cases

Case	Screening / NIPT / CVS / Amniocentesis	First-trimester USG	Detailed USG	Perinatal Outcome
1	High risk (Tr21 >1/50); NIPT: Tr16; CVS: Tr16 + inv(9)	No abnormal findings	Normal	IUGR, oligohydramnios, PROM; C/S at 34+6, 1450 g
2	NIPT: 45X/0; Amniocentesis: Normal karyotype	Single umbilical artery	Single umbilical artery	IUGR; C/S at 37+1, 1720 g
3	NIPT: Tr4 (high risk); no invasive test	No abnormal findings	Normal	Term C/S, 3150 g healthy
4	Screening: Tr21 risk 1/250; NIPT: Tr7; Amniocentesis: Normal karyotype	Mild bilateral pyelectasis + echogenic focus	Same findings	Term C/S, 3300 g healthy
5	Screening: Tr21 risk 1/477; NIPT: Tr7; Amniocentesis: Normal karyotype	No abnormal findings	Normal	Term delivery, 3190 g healthy
6	NIPT: Tr18; Amniocentesis: Normal karyotype	No abnormal findings	Normal	IUGR + preeclampsia; C/S at 38 wks, 2650 g
7	Screening: Tr21 risk 1/161; NIPT: 12p duplication; Amniocentesis: Normal karyotype	Hyperechogenic bowel ± foot deformity	Hyperechogenic bowel	Placental insufficiency + fetal distress; C/S at 34 wks, 2190 g

SS-015

Fetal Renal Füzyon anomalileri ve Perinatal Sonuçları

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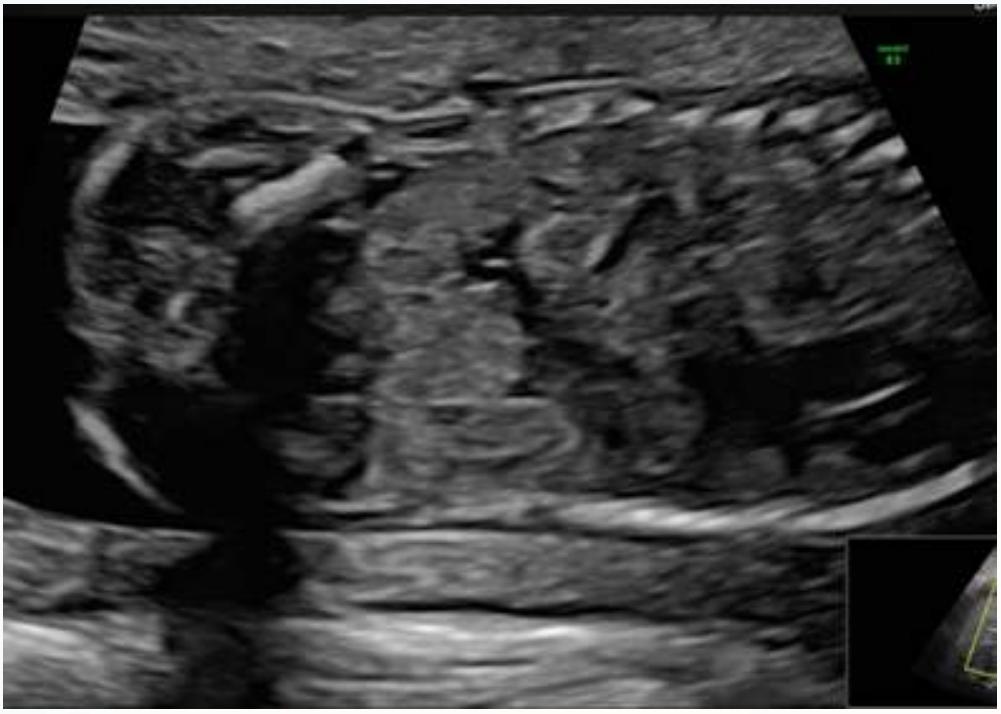
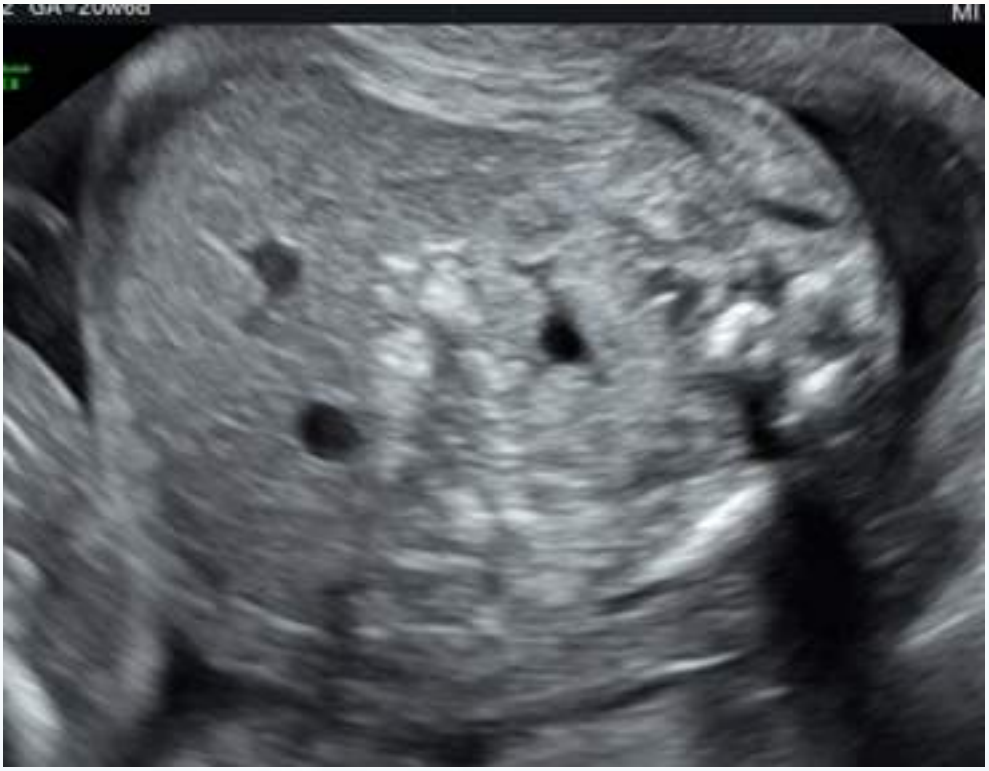
AMAÇ: Prenatal dönemde fetal renal füzyon anomalilerinin perinatal sonuçların değerlendirilmesi amaçlandı.
GEREÇ-YÖNTEM: 2023 Ocak-2025 Eylül tarihleri arasında perinatoloji kliniğine başvuran 18 olgu retrospektif değerlendirildi. Ultrasonografi muayenesinde tanı konulan olgulara prenatal genetik incelemede sitogenetik, array ve tüm ekzom analiz yapıldı. Postnatal dönemde Pediatrik Üroloji konsültasyonu planlandı.
BULGULAR: Değerlendirilen 18 olguya ortalama 21. gebelik haftasında tanı koyuldu. Olguların %67'sinde at nalı böbrek (n=12), %33'ünde cross fused böbrek (n=6) saptandı. Tüm olguların %11'inde hidroüreter bulunmakta olup (n=2), hepsi cross fused böbrek grubunda yer aldı. Tüm olgularda perine anatomisi normal değerlendirildi. %78 olguya ek majör sistem anomalisi eşlik etmekteydi (n=14). Cross fused böbrek nedeni ile takip edilen olguların %50'sinde (n=3); at nalı böbrek saptananların ise %92'sinde ek majör sistem anomalisi mevcuttu. Bu olgulara en sık kardiyovasküler anomaliler eşlik etti (n=11). Olguların %72'sinde amniyotik mayi normaldi. %78 hasta prenatal genetik analiz yaptırmış olup (n=14); bu hastaların beşinde genetik anomali saptadı. Tespit edilen genetik anomalilerin hepsi Trizomi 18 olarak sonuçlandı. Bu olguların dördü at nalı böbrek; biri ise cross fused böbrek grubunda bulunmaktaydı. Olguların %56'sı terminasyon (n=10), %17'si canlı doğum (n=3) ve %11'i ölü doğum (n=2) ile sonuçlandı. Prenatal dönemde cross fused olarak değerlendirilen ve canlı doğan iki olgudan birinin tanısı postnatal dönemde doğrulandı. Diğer canlı doğan olgu ise postnatal dönemde at nalı böbrek tanısı aldı. Üç hastanın gebelik takibine devam edilmektedir.
SONUÇ: Perinatal dönemde renal füzyon anomalileri, ek majör yapısal anomali ve kromozom anomalisi açısından ayrıntılı değerlendirilmelidir. Eşlik eden kromozom veya yapısal anomali varlığında prognoz kötü seyredebilmektedir. Prenatal genetik analiz tüm olgulara önerilmelidir.

Anahtar Kelimeler: At nalı, Cross fused, füzyon

At Nalı Böbrek



Cross Fused Böbrek



SS-016

Short term surgical treatment outcomes of cesarean scar pregnancies

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In this study, we aimed to retrospectively evaluate the cases of scar pregnancy diagnosed in the first trimester and treated with surgical methods in our clinic to determine the risk factors, early term results and early complications.

The gestational week, type of scar pregnancy, complications occurring during the surgical procedure and in the postoperative period, blood transfusion requirement, intensive care requirement and hospitalization days in 85 cases and 85 control group cases who underwent surgical treatment with the diagnosis of scar pregnancy in the first trimester between 01.01.2010 and 31.12.2023 in our clinic were evaluated retrospectively by examining hospital records.

Surgical treatment complication rate was higher in scar pregnancy cases (40% and 3.5%) compared to the control group ($p<0.001$). Bleeding (23.5% and 3.6%) and foley balloon application (27.1% and 2.4%) were observed at a higher rate in the scar pregnancy group ($p<0.001$, $p = 0.028$; $p<0.001$). The rate of complications in type 2 scar pregnancies (62.1%) was higher than in type 1 scar pregnancies (28.6%) ($p<0.001$). Hematoma (51.7% and 19.6%), foley balloon (44.8% and 17.9%) and tranexamic acid injection (31% and 8.9%) were observed at higher rates ($p<0.001$). It was revealed that complications were more severe in scar pregnancy cases diagnosed in advanced weeks. Surgical treatment is more successful and complication rate is low in cases of scar pregnancy diagnosed early. Early diagnosis and treatment of scar pregnancies is a preventive medicine practice that will reduce mortality and morbidity due to placenta acreata spectrum in the later weeks of pregnancy.

Keywords: Cesarean scar pregnancy, curettage, ectopic pregnancy.

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SS-017

Cytomegalovirus Screening During Pregnancy: Two-Year Experience of a Tertiary Care Centre

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OBJECTIVE: Congenital cytomegalovirus (CMV) is the most common congenital infection and an important cause of neonatal morbidity. While some countries perform routine screening, national guidelines in Turkey do not recommend it. This study evaluated CMV IgM testing at a tertiary care center to assess the necessity of routine screening during pregnancy.

Material and METHODS: Pregnant women who underwent CMV serology at Kastamonu Training and Research Hospital between April 2023 and May 2025 were retrospectively analyzed. For all IgM-positive cases, IgG and avidity results and, when available, amniocentesis CMV PCR findings were recorded. Postnatal outcomes were evaluated.

RESULTS: A total of 2,690 women were tested. Forty-one underwent testing due to ultrasound findings such as fetal growth restriction, discordant biometry, or hyperechogenic bowel; none were IgM positive. Among 2,649 women screened routinely, 101 (3.7%) were IgM positive. Ninety-two had high-avidity IgG, indicating past infection, and nine showed low avidity, consistent with acute infection. None of these patients consented to amniocentesis. Fetal growth restriction developed in five acute cases. Postnatal follow-up revealed six infants with normal development, one with thyroid agenesis, one neonatal cardiac death, and one elective termination due to aortic coarctation. None of these were attributable to congenital CMV.

CONCLUSION: Screening identified very few acute infections, and numerous tests were required to determine clinically significant results. Given the limited sample size, the relative effectiveness of routine screening versus targeted screening cannot be determined. Larger population-based studies are needed to inform optimal screening policies.

Keywords: Cytomegalovirus, Pregnancy, Serology

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SS-018

Late preterm pregnancy with phytobezoar-related mechanical intestinal obstruction: A case report

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OBJECTIVE: A bezoar is a mass formed by indigestible materials in the digestive system. Reports of bezoars during pregnancy are limited, with most involving trichobezoars. Phytobezoar cases are exceptionally rare; only one case report and one poster presentation were found in databases, both in patients with a history of bariatric surgery. Our case is the first reported phytobezoar in a pregnant woman without prior bariatric surgery. Therefore, this case aims to contribute to the literature on the management of phytobezoar-induced gastrointestinal obstruction during pregnancy.

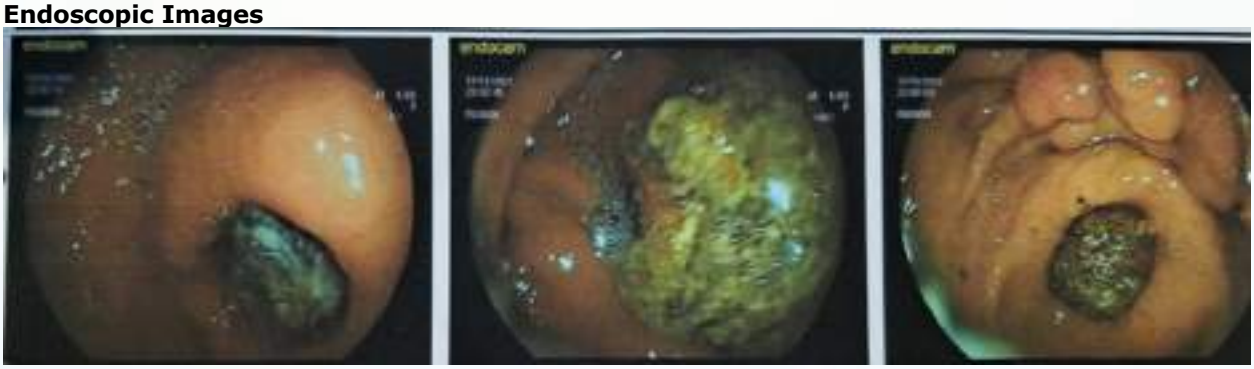
CASE: A 38-year-old G3P2 patient at 35 weeks of gestation, with no additional medical history and two previous cesarean sections, was admitted due to abdominal pain, nausea, and vomiting. Abdominal ultrasound revealed a 47 mm echogenic bezoar at the gastric outlet. Obstetric ultrasound showed adequate amniotic fluid and a reactive non-stress test without contractions. Upper endoscopy identified a bezoar covering the bulbus and pylorus, which was fragmented but not fully removed. General surgery recommended conservative management with cola and pineapple juice. Due to worsening oral intake, total parenteral nutrition was initiated. At 36 weeks, due to persistent symptoms, a cesarean was performed via Pfannenstiel incision, delivering a healthy male infant (2700 g, Apgar 9–10). Through the same incision, a 6x4x4 cm phytobezoar was removed via enterotomy and primarily repaired. The patient tolerated oral intake by postoperative day 3 and was discharged without complications.

CONCLUSION: A multidisciplinary approach is essential to minimize complications in the diagnosis and management of mechanical intestinal obstruction due to phytobezoar during pregnancy.

Keywords: Mechanical intestinal obstruction, phytobezoar, pregnancy

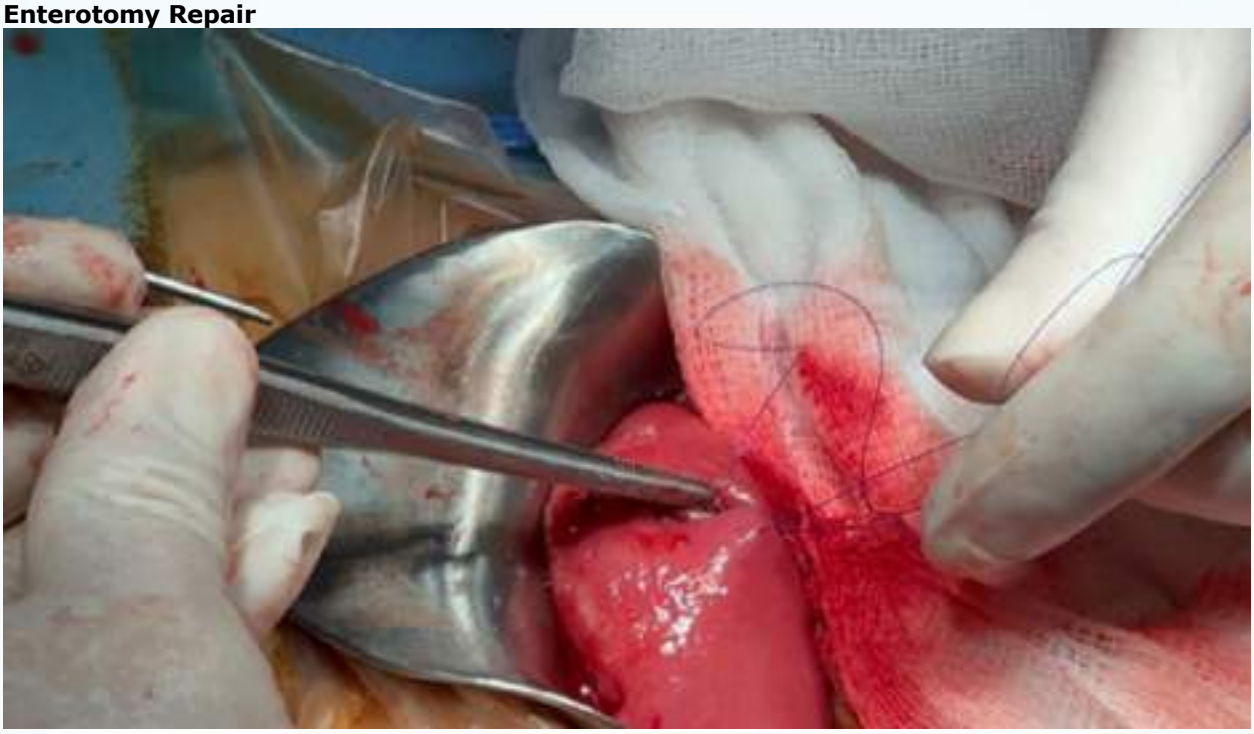
6x4x4 cm. Measuring Phytobezoar

Endoscopic Images



a-Before procedure b-Broken Piece c-After Procedure

Enterotomy Repair



Removal of the Bezoar

SS-019

Congenital diaphragmatic hernia: A 10 year single center experience

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INTRODUCTION: Congenital diaphragmatic hernia (CDH) is a rare defect from incomplete diaphragm formation, causing abdominal organs to herniate into the thorax. It results in pulmonary hypoplasia, pulmonary hypertension, and high mortality.

METHOD: This retrospective study analyzed the amniocentesis results, LHR values and neonatal outcomes of 28 fetuses with CDH managed at our clinic between 2016 and 2025.

RESULTS: The amniocentesis results, LHR values, and neonatal outcomes of 28 fetuses with CDH diagnosed were evaluated. Of the three fetuses undergoing amniocentesis, one had trisomy 13, one had trisomy 18, one had pericentric inversion on chromosome 9. Pregnancy termination was performed, while nine fetuses underwent TOP due to family reasons. Neonatal survival was 44%. As reported in literature, low LHR values predicted poor survival. When the neonatal results of the remaining 16 fetuses were evaluated, it was observed that the LHR results in their prenatal follow-ups were compatible. While all of the fetuses with an LHR value below 1 resulted in postnatal death, only 1 of the 8 fetuses with values above 1.4 resulted in postnatal death. The analysis shows that LHR is a strong determinant of survival. At low LHR values (below ~1.0), the probability of survival is nearly zero, but it rises sharply between 1.3–1.5, reaching almost 100% at LHR ≥ 2.0. This indicates a clear threshold effect: higher LHR values correspond to significantly greater survival likelihood, while low values strongly predict an "Ex" outcome.

CONCLUSION: Prenatal LHR assessment and detection of anomalies are key for prognosis and counseling.

Keywords: Congenital diaphragmatic hernia, LHR, prognosis

SS-020

Congenital Diaphragmatic Hernia with Isolated Bowel Herniation in Both Fetuses of a Monochorionic Diamniotic Twin Pregnancy: The Role of Doppler Ultrasound in Prenatal Diagnosis

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Zeynep kamil kadın ve çocuk hastalıkları eğitim ve araştırma hastanesi, Perinatoloji kliniği

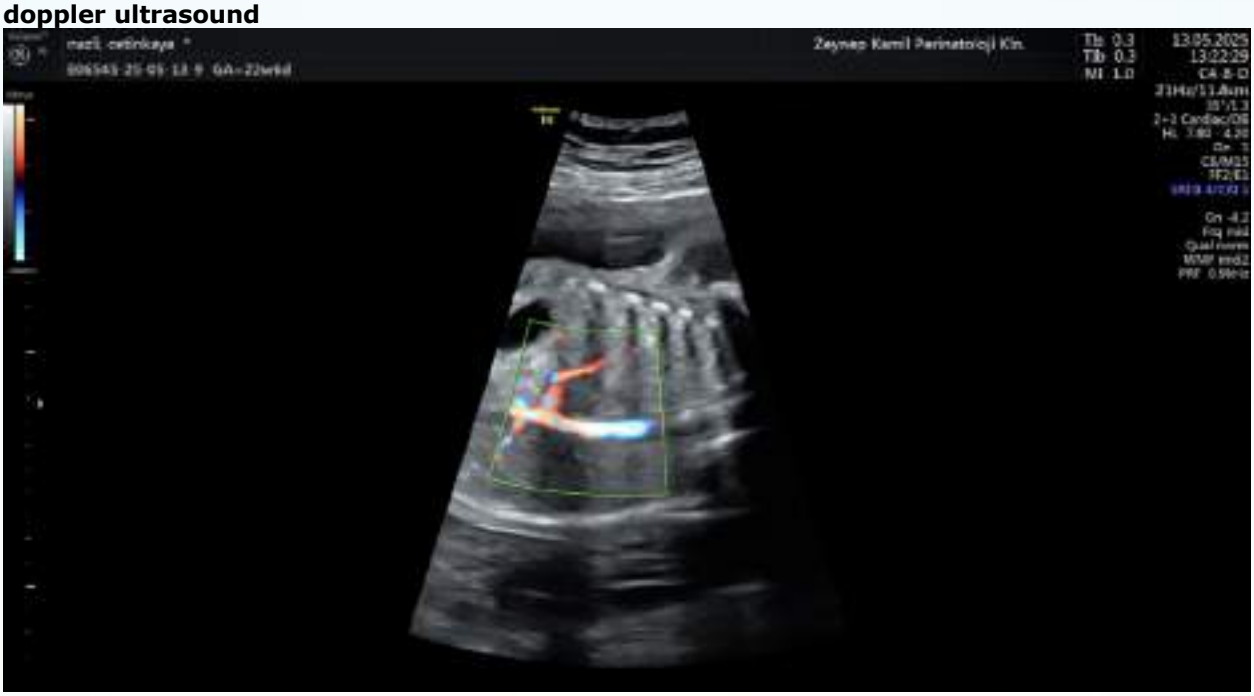
INTRODUCTION: Congenital diaphragmatic hernia (CDH) is a developmental defect of the diaphragm, allowing abdominal organs to herniate into the thoracic cavity. It occurs in approximately 1 in 3000 live births and is associated with high neonatal mortality. We report a rare case of a monochorionic diamniotic (MCDA) twin pregnancy in which both fetuses presented with left-sided CDH showing the "isolated bowel" phenotype, with intra-abdominal stomachs.

CASE: A 31-year-old G2P1 woman with a previous cesarean was referred at 18 weeks due to rightward cardiac displacement in one fetus, initially suspected as microcystic CPAM. Amniocentesis was performed for both fetuses at 20 weeks. At 22 weeks, the stomach and liver were confirmed intra-abdominal, but Doppler ultrasound revealed the superior mesenteric artery coursing into the thoracic cavity, establishing the diagnosis of left CDH. The co-twin initially showed no abnormality; however, at 27 weeks, similar left-sided CDH with isolated bowel herniation and peristalsis displacing the heart to the right hemithorax was detected. Genetic testing, including clinical exome sequencing, revealed no pathogenic variants. At 33 weeks, spontaneous preterm labor occurred, and both neonates were delivered via cesarean section. Surgical repair on day two confirmed the intra-abdominal position of the stomachs. Postnatal course has been uneventful to date.

DISCUSSION: Although CDH in both twins has been previously reported, this is the first documented case of bilateral isolated bowel-type CDH in an MCDA pregnancy. This case emphasizes the value of detailed Doppler assessment, particularly of the mesenteric vessels, in differentiating CDH from other thoracic anomalies during early gestation.

Anahtar Kelimeler: congenital diaphragmatic hernia,monochorionic diamniotic twin pregnancy, Doppler ultrasound

doppler ultrasound



doppler ultrasound of superior mesenteric arter at 22 gw

SS-021

A case of placental chorioangioma treated with microwave ablation under ultrasound guidance

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INTRODUCTION: Placental chorioangioma is a mass composed of capillary vascular structures. It occurs in approximately 0.5–1% of. The perinatal mortality rate for large masses is about 28%. Therefore, both the size of the mass and the degree of vascularity are decisive for prognosis.

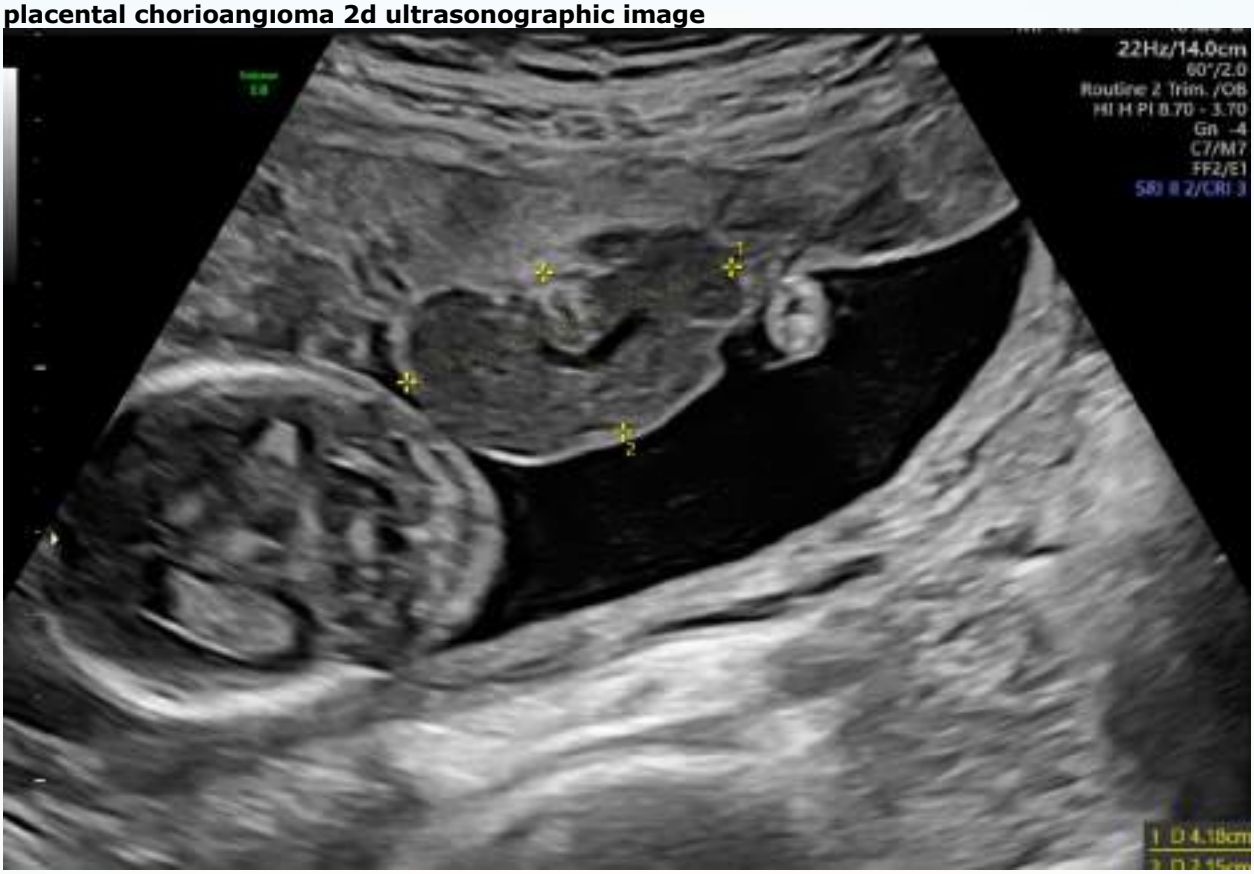
Material and METHODS: A case of placental chorioangioma treated with microwave ablation under ultrasound guidance is presented.

Case Presentation: A 31-year-old, gravida 1, para 0 woman with a 17-week-and-5-day pregnancy was evaluated at the Perinatology Clinic due to elevated AFP levels. Placental assessment revealed a 41 × 23 mm area with increased vascularity, bulging toward the amniotic cavity, localized to the right side near the fetal umbilical cord insertion, with hypoechoic areas and a feeding vessel measuring 3 mm at its widest point, consistent with a chorioangioma. The case was monitored for possible complications. At the 19-week-and-5-day follow-up, the chorioangioma measured 51 × 40 mm. MCA PSV was 40.4 cm/s. Given that the dimensions exceeded 5 cm, intense vascularization was observed, and fetal anemia was detected, the patient was informed about treatment options. At 20 weeks and 5 days, the patient consented to chorioangioma devascularization, and the procedure was performed with MCA PSV at 46 cm/s Under ultrasound guidance, ablation of the feeding vessel was performed using microwaves. No blood flow was observed on control sonography. The procedure was successful.

Conclusion: Treatment options are considered when complications develop. Microwave ablation may be considered as a treatment option to eliminate the shunt effect.

Keywords: chorioangioma, microwave ablation, fetal anemia

placental chorioangıoma 2d ultrasonographic image



SS-022

Prenatal Dönemde Saptanan Umbilikal Kord Kistleri: Klinik Özellikleri ve Perinatal Sonuçları

Cagdas Nurettin Emeklioglu, Münip Akalın, Esra Esim Buyukbayrak
Marmara Üniversitesi Pendik Eğitim Araştırma Hastanesi, Kadın Hastalıkları ve Doğum ABD, Perinatoloji BD

Giriş: Umbilikal kord kistleri, prenatal ultrasonografi sırasında insidental olarak saptanan nadir anomalilerdir ve gebeliklerin %0,4–3,4'ünde bildirilmiştir. Yüksek çözünürlüklü ultrasonografinin yaygınlaşması, bu kistlerin daha sık saptanmasını sağlamış, ancak beraber yönetim konusundaki belirsizlik tam anlamıyla ortadan kaldırılamamıştır. Bu çalışmada amacımız, kliniğimizde umbilikal kord kisti saptanan 7 olgunun klinik özelliklerini ve perinatal sonuçlarını literatür ışığında sunmaktır.

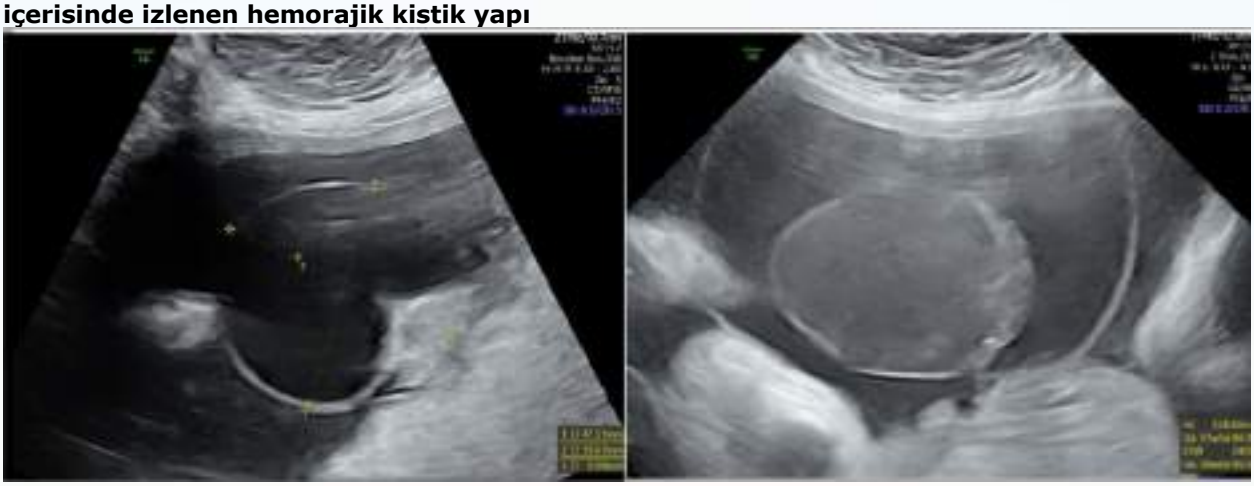
Materyal ve Metot: Ocak 2022–Aralık 2024 arasında üçüncü basamak merkezimize başvuran ve ultrason ile ≥3 mm anekoik umblikal kord kisti saptanan gebeler çalışmaya dahil edildi. Demografik veriler, kist özellikleri, eşlik eden anomaliler ve perinatal sonuçlar kayıt edilip tanımlayıcı istatistiklerle analiz edildi.

Bulgular: Toplam 7 olgu saptandı; ortalama maternal yaş 30,8, tanı haftası 23'tü. İki olgu ilk, üç olgu ikinci trimesterde saptandı. Altı olguda kist persiste ederken bir olgu spontan regresyon göstermiştir. Vaka serimizde kist çapları 13*14 mm ile 90*60 mm arasında olup, saptanmış en büyük kistte takiplerde kist içeriğinde hemorajik kistik bir oluşumun meydana geldiği gözlenmiştir. İki olguda umblikal kord kistine majör fetal anomaliler eşlik etmekteydi; bu vakaların birinde aile terminasyon seçeneğini tercih etti, diğerinde ise takipte erken başlangıçlı geli-şim geriliği nedeniyle 33. Gebelik haftasında sezaryen ile doğum gerçekleştirilmiştir. Bir vaka TTTS evre 4 olup takipten çıkmış ve perinatal sonucu öğrenilememiştir. İzole kord kisti olan dört olguda term doğum ve sağlıklı neonatal sonuçlar elde edilmiştir.

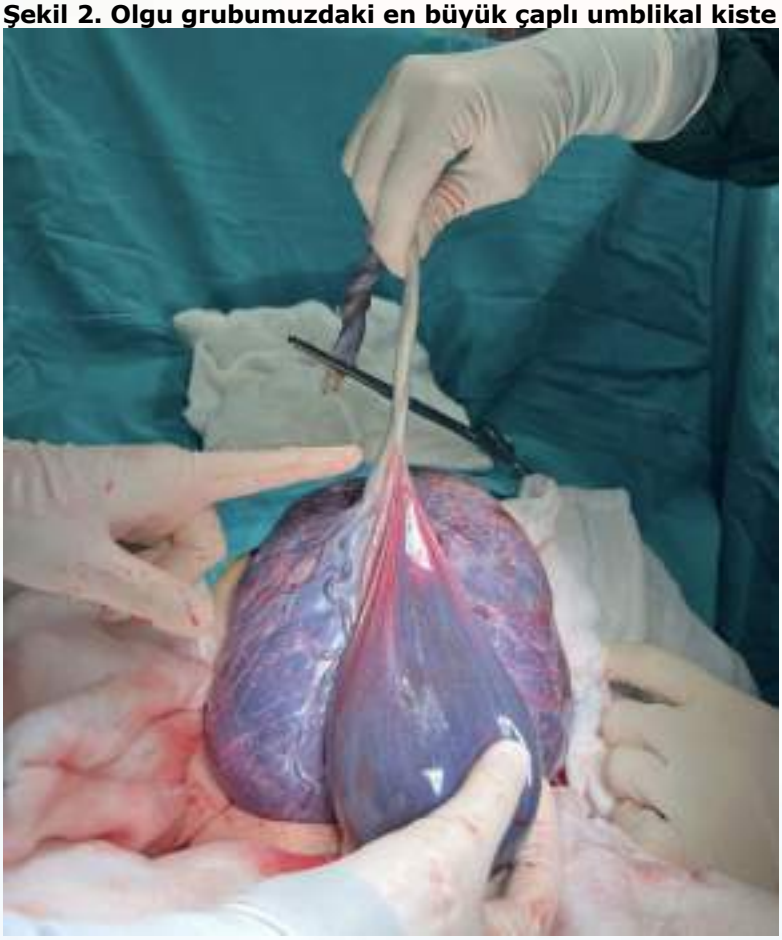
Sonuç: İzole kord kistleri genellikle iyi prognozludur; ancak eşlik eden anomali veya gelişim geriliğinde aileye ayrıntılı danışmanlık ve invaziv testlerin gerekli olmaktadır. Umblikal kord kistleri masum bir ultrason bulgusu olarak değerlendirilmemeli, ek fetal anomaliler açısından detaylı fetal anatomik tarama yapılmalıdır.

Anahtar Kelimeler: umblikal kord kisti, fetal anomali, perinatal sonuçlar

Şekil 1. Olgu grubumuzdaki en büyük çaplı umblikal kist ve takiplerde ortaya çıkan kist içerisinde izlenen hemorajik kistik yapı



Şekil 2. Olgu grubumuzdaki en büyük çaplı umblikal kiste ait peroperatif görsel



Tablo 1. Umblikal kord kisti saptanan olguların özellikleri ve perinatal sonuçları

Olgu	Gebelik Haftası	Anne Yaşı	Persistans	Karyotip	Ek Ultrasonografik Bulgu	Kist	Doğum	Doğum Haftası	Doğum Kilosu	APGAR	YDYBÜ
1	38	29	Evet	Yok	Yok	34*40	C/S	39	3700	9-10	Yok
2	23	35	Evet	Yok	MKDA, Evre 4 TTTS, Fet1: DV ters A, TR pansistolik	16*14	C/S	-	-	-	-
3	31	33	Evet	Yok	Yok	48*50	NSD	37	3075	8-9	Yok
4	12	24	Hayır	Yok	Yok	24*30	NSD	40	3650	8-9	Yok
5	12	33	Evet	Normal	Oligohidroamnios + Kardiyak hipertrofi + Bilateral multikistik displas-tik böbrek + PRUV + DV agenezisi + Tek umblikal arter + Umblikal kord kisti + Ambigus genitalia	14*13	Terminasyon	-	-	-	-
6	23	27	Evet	Yok	Unilateral multikistik displastik böbrek + Erken başlangıçlı FGR + Sağ ventrikül hipertrofisi	27*12	C/S	33	1335	5-7	Var
7	22	35	Evet	Yok	Yok	90*60	C/S	38	3345	7-9	Yok

SS-023

Prenatal Diagnosis of Sotos Syndrome with Concurrent NSD1 and WDR11 Mutations: A Rare Case Report

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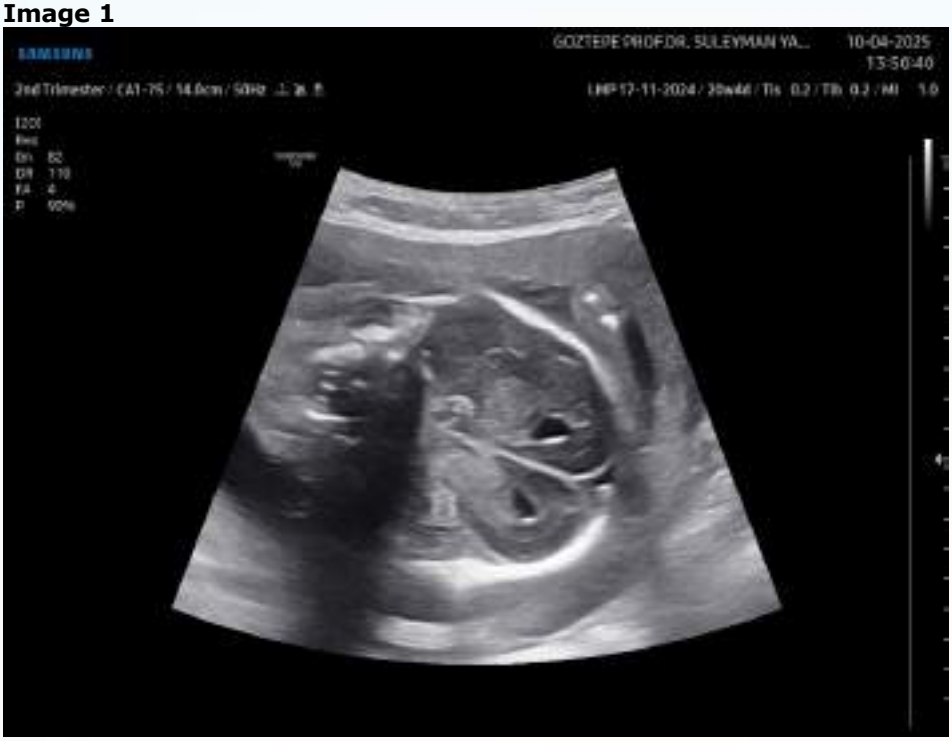
İstanbul Medeniyet Üniversitesi, Göztepe Prof. Dr. Süleyman Yalçın Şehir Hastanesi

Sotos syndrome is a rare overgrowth disorder with autosomal dominant inheritance,typically associated with mutations or deletions in the NSD1 gene.Prenatal diagnosis is challenging due to the absence of specific ultrasound findings.In recent years,whole-exome sequencing(WES) has made a significant contribution to prenatal diagnosis.Here,we present a case of prenatally diagnosed Sotos syndrome. In the ultrasonography performed on the 12+3weeks pregnant woman who applied for the first trimester screening test,nuchal translucency was measured as 4.95mm.Genetic consultation was done.The family opted for chorionic villus sampling(CVS).Conventional karyotyping,microarray and FISH analysis revealed no abnormalities.At 20+4weeks,second trimester ultrasonography showed that the frontal horns of the lateral ventricles were square and the fourth ventricle was dysmorphic.(Image-1)Genetic counseling was provided and amniocentesis with WES was recommended. At 28weeks,ultrasound revealed delayed sulcation of the insula,parieto-occipital sulcus,and calcarine sulcus,with absence of the cingulate gyrus.(Image:2-3)Fetal cranial MRI reported that gyrus development was retarded according to the week.As a result of WES,mutations were detected in the NSD1gene that causes Sotos syndrome and in the WDR11gene in addition.The findings were confirmed by Sanger analysis.

This case highlights the predictive value of increased nuchal translucency in the first trimester for genetic syndromes.It also emphasizes that cortical malformations may become apparent later,even if earlier neurosonography appears normal.Careful evaluation of ultrasound and MRI findings,alongside advanced genetic testing such as WES,is crucial for prenatal diagnosis.This case is a rare prenatal diagnosis example in the literature in terms of detecting WDR11mutation along with NSD1mutation,and reveals the importance of a multidisciplinary approach and providing the right counseling to the family.

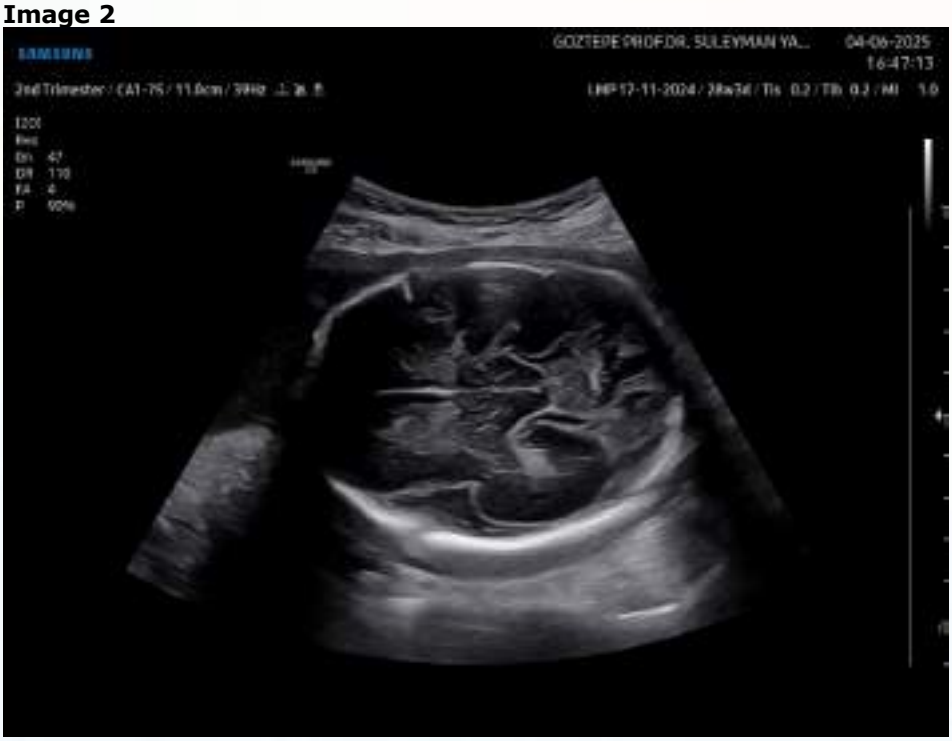
Keywords: Sotos syndrome, NSD1 gene, WDR11 gene, Whole-exome sequencing (WES)

Image 1



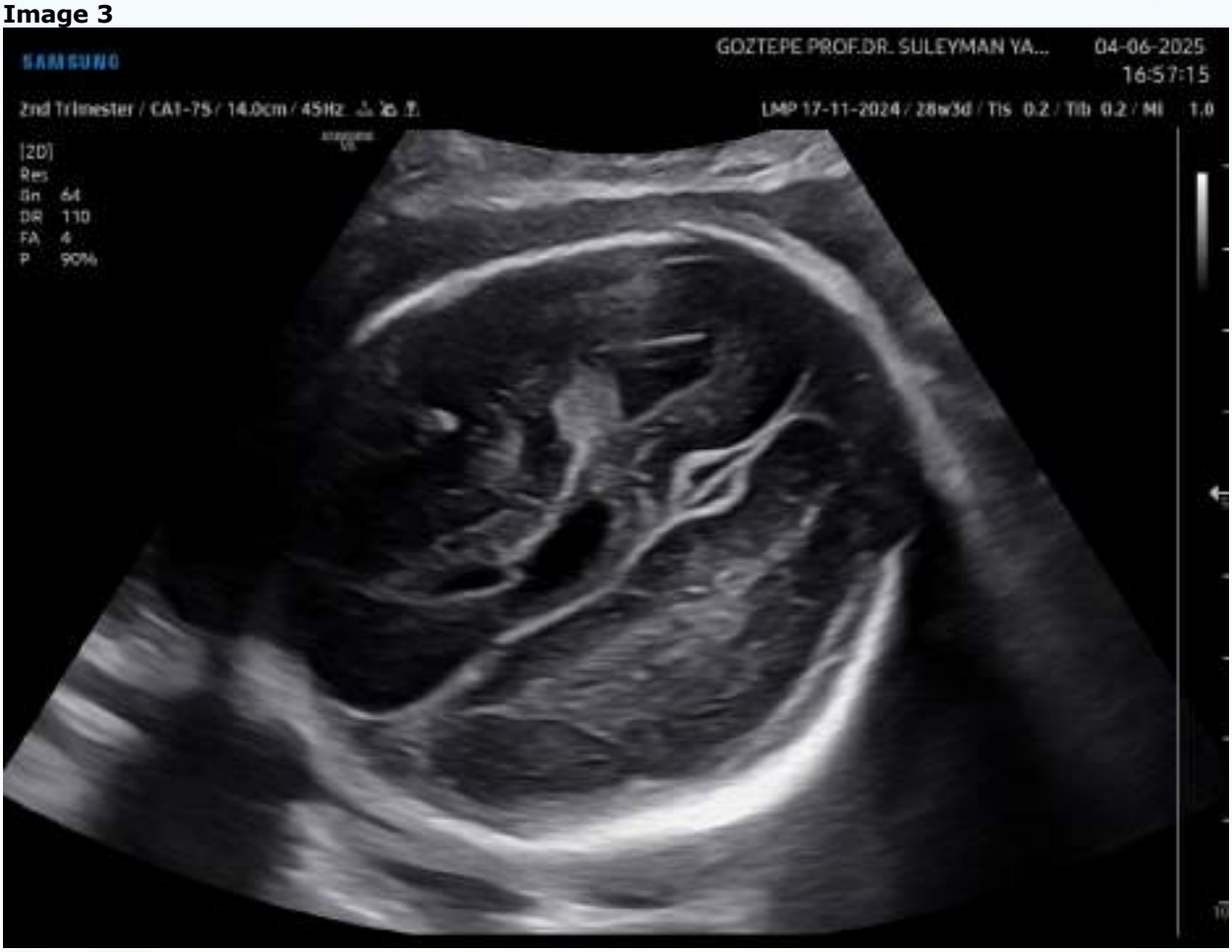
The frontal horns of the lateral ventricles were square

Image 2



Delayed sulcation of the insula

Image 3



Delayed sulcation of the parieto-occipital sulcus

SS-024

Prenatal Diagnosis and Postnatal Management of Polymelia: A Rare Case Report

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INTRODUCTION: Polymelia is a rare congenital anomaly characterized by the presence of supernumerary limbs, with an estimated incidence of 6 per 10,000 live births. Its etiology is multifactorial, involving genetic, embryological, and environmental factors.

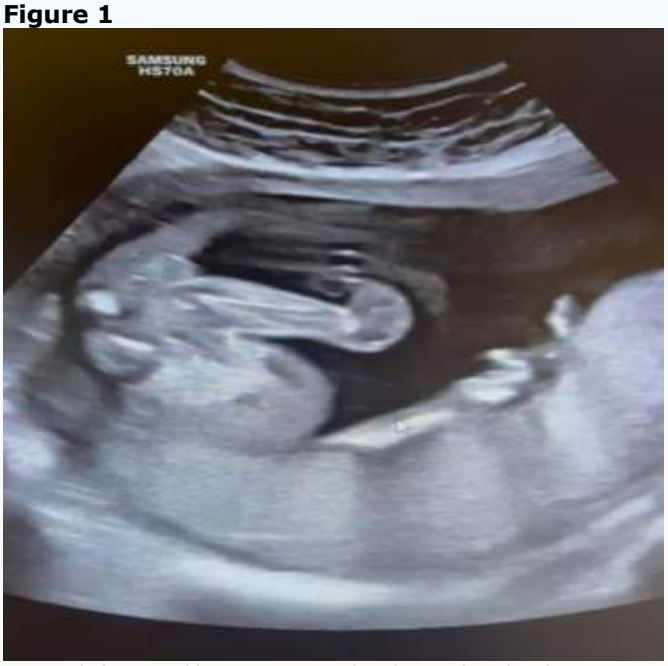
CASE REPORT: A 30-year-old gravida 4 woman was referred at 19 weeks of gestation for suspected clubfoot. Detailed ultrasonography revealed normal fetal biometry and amniotic fluid volume. In addition to two normal lower limbs, a third limb arose from the sacrococcygeal region, fixed in a flexed position. Intracranial, cardiac, abdominal, and genitourinary anatomy were unremarkable, and the fetus was female. Amniocentesis showed a normal 46,XX karyotype. Pregnancy progressed uneventfully, and an elective cesarean section at 39 weeks delivered a 3200 g female infant (Apgar 9/10). Postnatal examination confirmed a third limb and hemipelvis attached to the sacrum without active motion. MRI revealed a 5 × 3 cm heterogeneous lesion at the sacral level, consistent with myelomeningocele. At six months, surgical excision of the accessory limb and pelvic bones was performed, with uneventful recovery. At two years, the child exhibited normal growth, continence, and motor milestones.

DISCUSSION: Polymelia likely arises from disruption of limb bud development during the 4th–5th embryonic week, possibly due to aberrant fibroblast growth factor signaling or incomplete twinning. In this case, absence of teratogenic or hereditary factors suggests a sporadic cause.

CONCLUSION: This case highlights the value of prenatal imaging for early diagnosis and the importance of multidisciplinary management to ensure optimal surgical and developmental outcomes.

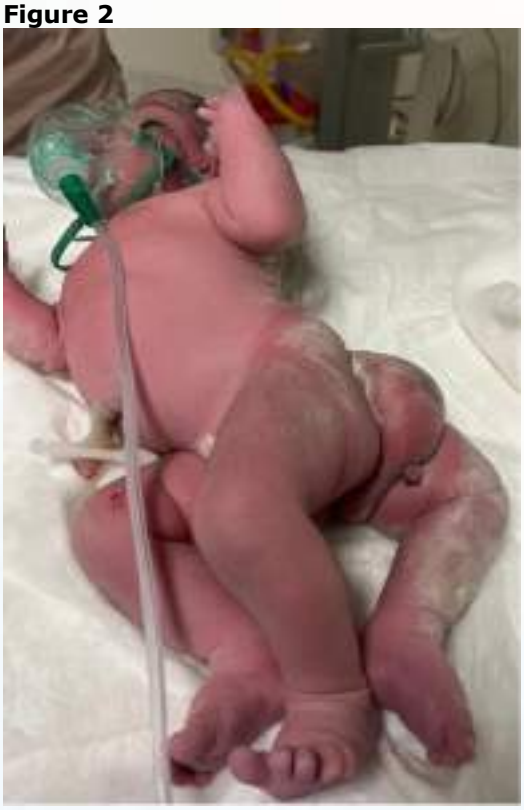
Keywords: Accessory limb, congenital anomaly, limb development, myelomeningocele, polymelia, prenatal diagnosis

Figure 1



Prenatal ultrasound image at 19 weeks of gestation showing a supernumerary third lower limb originating from the sacrococcygeal region. The accessory limb appears fixed in a flexed knee position and lacks movement.

Figure 2



Postnatal image of the newborn with a supernumerary third lower limb originating from the sacrococcygeal region.

SS-025

Prenatal diagnosis of fetal megalourethra with spontaneous resolution: report of two cases

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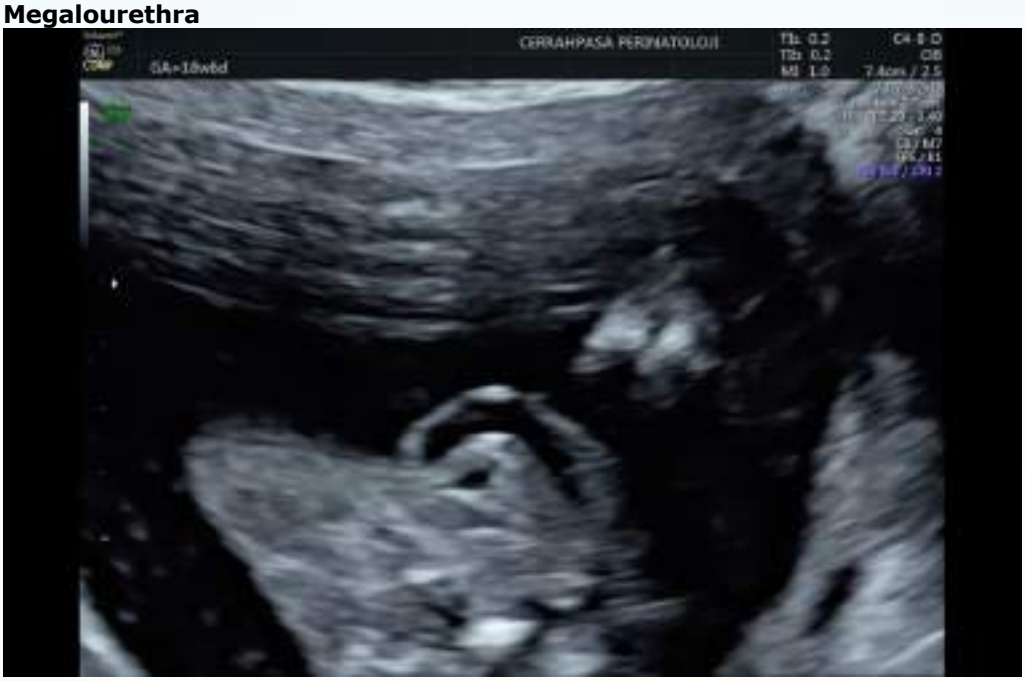
BACKGROUND: Congenital megalourethra is a rare anomaly characterized by abnormal dilatation of the penile urethra, often associated with lower urinary tract obstruction and poor prognosis. Most reported cases are complicated by renal impairment, pulmonary hypoplasia, or perinatal mortality. Nevertheless, spontaneous resolution during gestation has occasionally been described.

Cases: We present two fetuses diagnosed with prenatal megalourethra and suspected lower urinary tract obstruction. Both cases were detected during routine second-trimester ultrasonography and showed enlarged bladder and enlarged penile urethra. Chromosomal abnormalities were excluded by invasive procedures. Follow-up ultrasonography performed in the middle of pregnancy revealed that the previous abnormal findings had completely resolved in both cases. Outcome: Both pregnancies reached term without complications and resulted in the birth of healthy babies. No urinary system pathology was detected during postpartum follow-up.

CONCLUSION: These cases emphasize that, despite being commonly associated with poor outcomes, prenatal megalourethra can resolve spontaneously and may show a favorable neonatal prognosis. Recognition of this rare course is important for prenatal counseling and clinical decision-making. Our report contributes to the limited literature describing spontaneous resolution of fetal megalourethra.

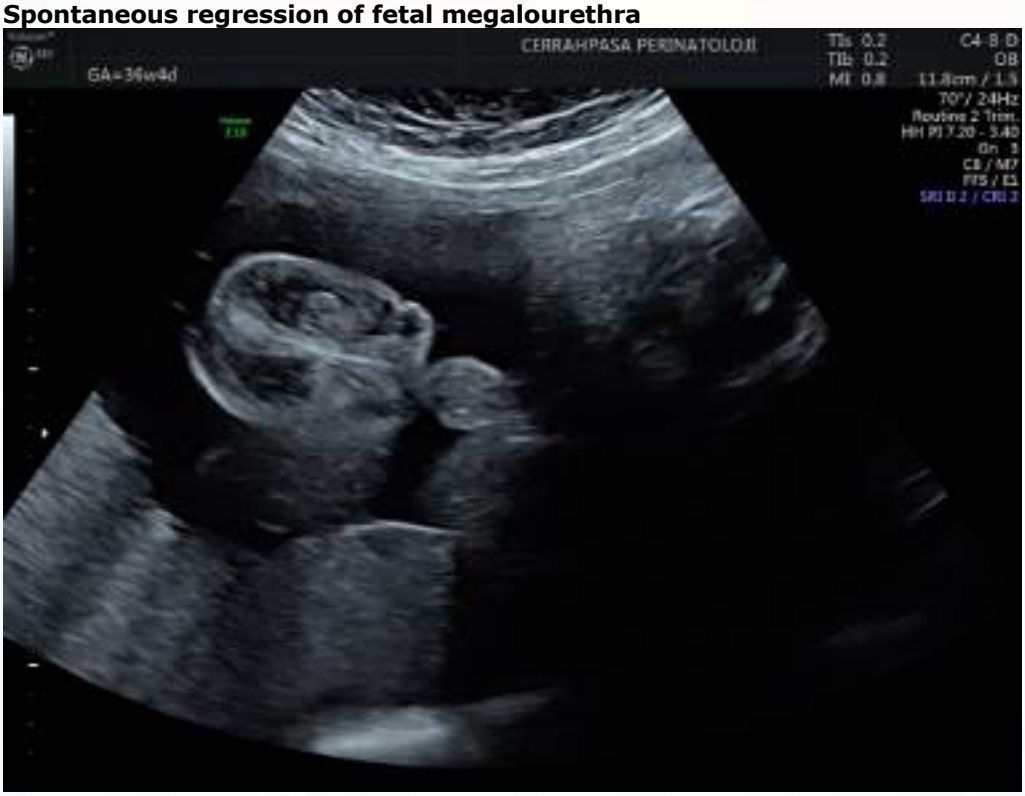
Keywords: bladder, congenital megalourethra, prenatal diagnosis, prenatal ultrasonography

Megalourethra



Appearance of megalourethra at the time of diagnosis

Spontaneous regression of fetal megalourethra



Spontaneous regression in the following weeks

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SS-026

Rare Association of Fetal Ascites Due to Bladder Rupture with Fetal Posterior Urethral Valve: Case Report

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[2]Department of Perinatology, Osmaniye State Hospital, Osmaniye, Turkey

INTRODUCTION: Posterior urethral valve(PUV) is a congenital anomaly characterized by the formation of obstructive membranous folds. Although PUV is the most common cause of lower urinary tract obstruction in male fetuses, bladder rupture due to this is a rare complication. In the presented case, the PUV diagnosis was established at early gestational age(GA) and a vesicoamniotic shunt was placed; however the increased intraluminal pressure proximal to the obstruction resulted in bladder rupture and fetal ascites.

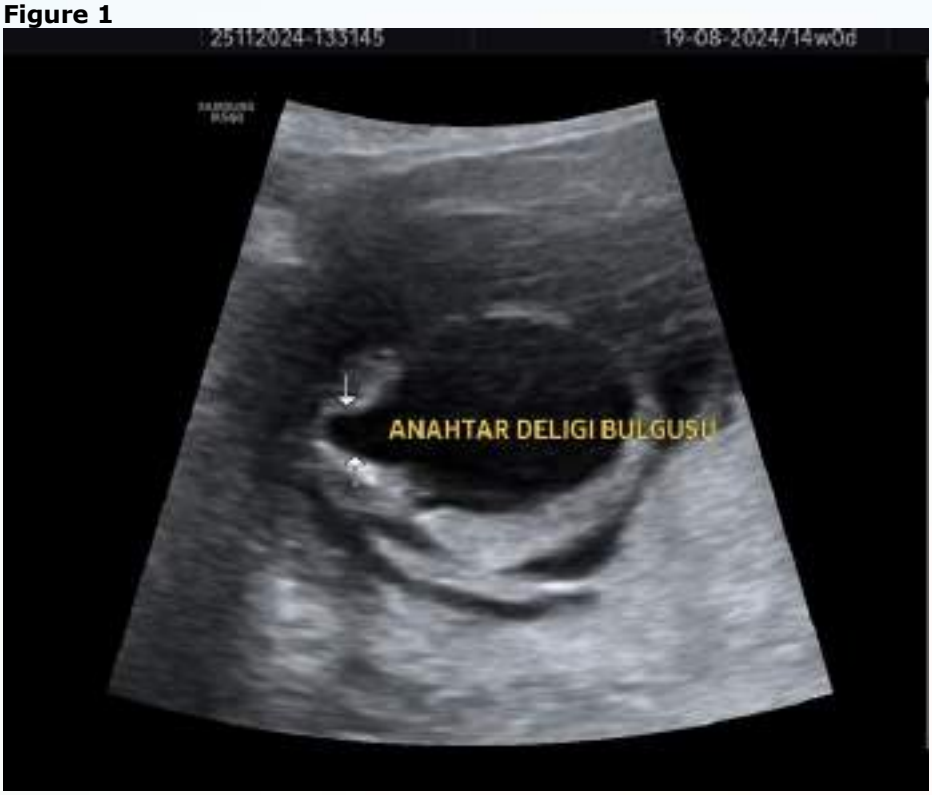
CASE REPORT: A 31 year old patient, G3P1A1L1, applied to our clinic at 14 GA. Ultrasonographic examination performed at this time revealed a distended bladder measuring 26x23mm accompained by the presence of the 'keyhole sign'(Figure 1,2,3).

The detailed fetal anatomical survey at the 18th GA following the vesicoamniotic shunt placement demonstrated oligohydramnios and intrabdominal free fluid(Figure 4). Applied CVS revealed normal karyotype and microarray. At 26 GA, the fetal abdominal circumference was significantly increased and fetal cardiac activity was absent. A stillborn 701 grams male fetus was delivered by cesarean section.

DISCUSSION: PUV can be identified on prenatal ultrasound through key findings such as megacystis, oligohydramnios and posterior urethral dilatation, often presenting as characteristic 'keyhole sign'. [1] In the present case, oligohydramnios observed at 18 GA indicated a poor prognosis, and despite the placement of a vesicoamniotic shunt, fetal demise couldn't be prevented. This case highlights the critical role of early prenatal ultrasonography in detecting PUV and predicting potential perinatal outcomes.

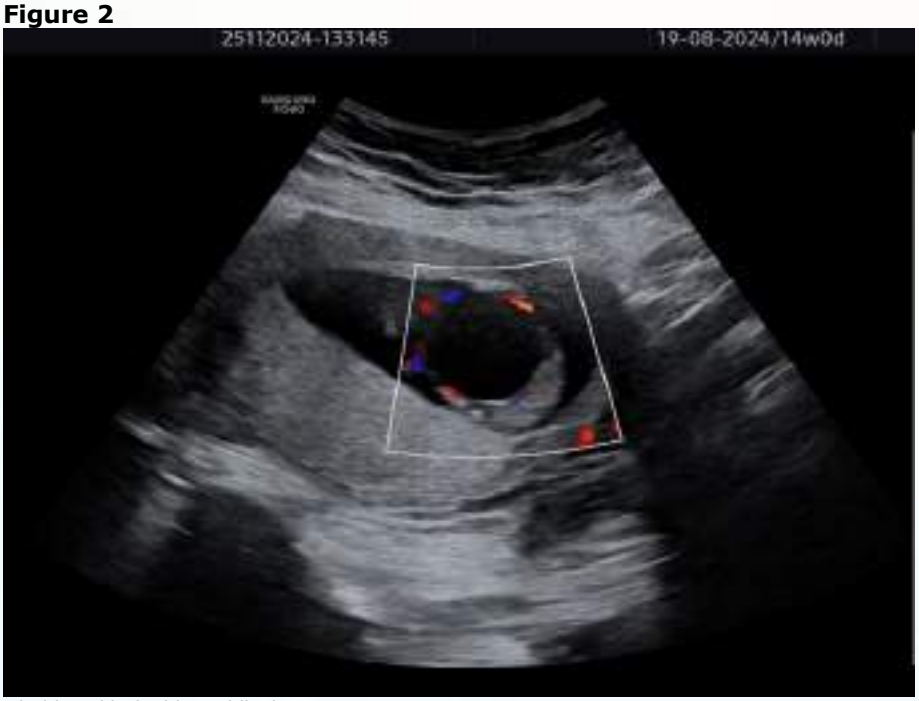
Keywords: Fetal ascites, Posterior urethral valve (PUV), Vesicoamniotic shunt

Figure 1



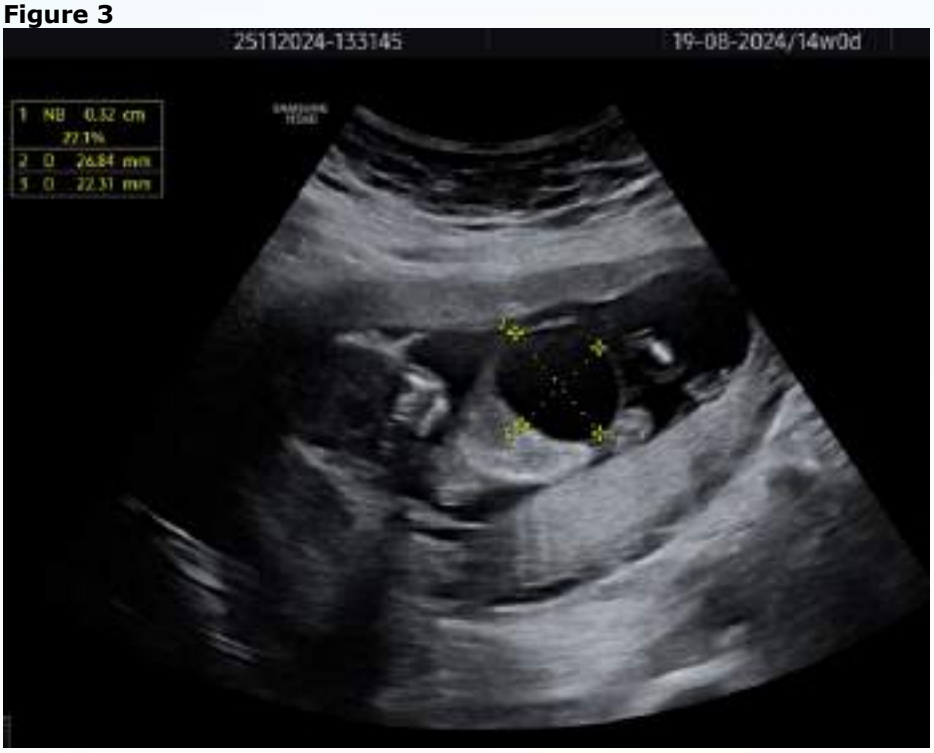
Keyhole Sign

Figure 2



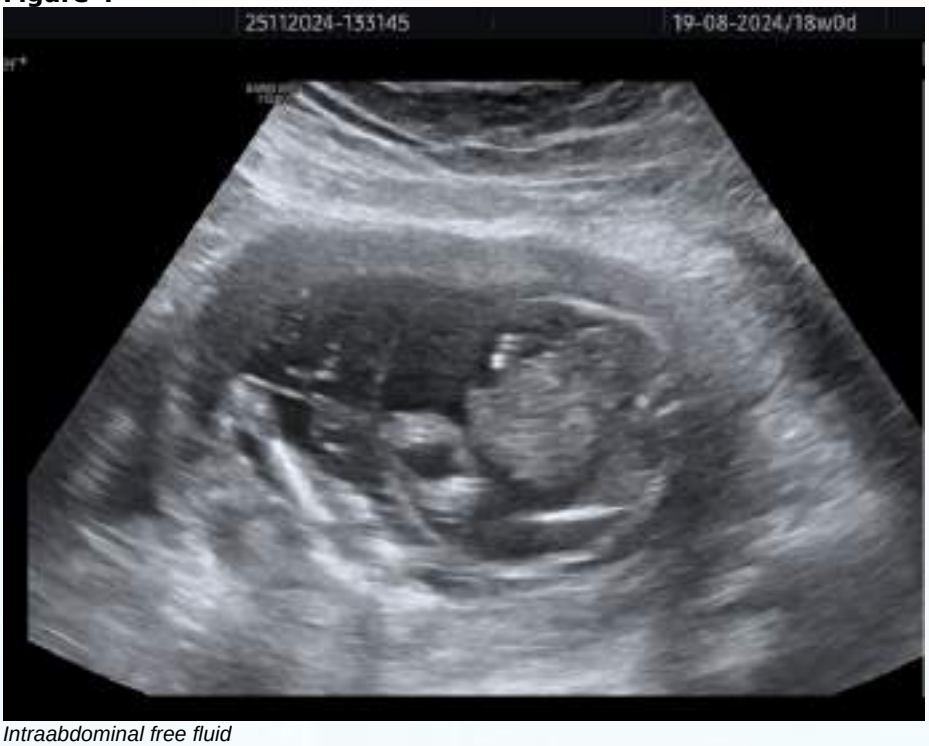
Bladder with double umblical arter

Figure 3



Bladder dimensions

Figure 4



Intraabdominal free fluid

SS-027

From Fetus to Father: A Rare Case of Prenatal CMT1A Diagnosis Leading to Familial Genetic Screening

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BACKGROUND: Charcot–Marie–Tooth disease type 1A (CMT1A), caused by a PMP22 gene duplication, is the most common hereditary neuropathy. Prenatal diagnosis is exceedingly rare due to its typically late clinical onset, and is often incidental during investigations for other fetal abnormalities.

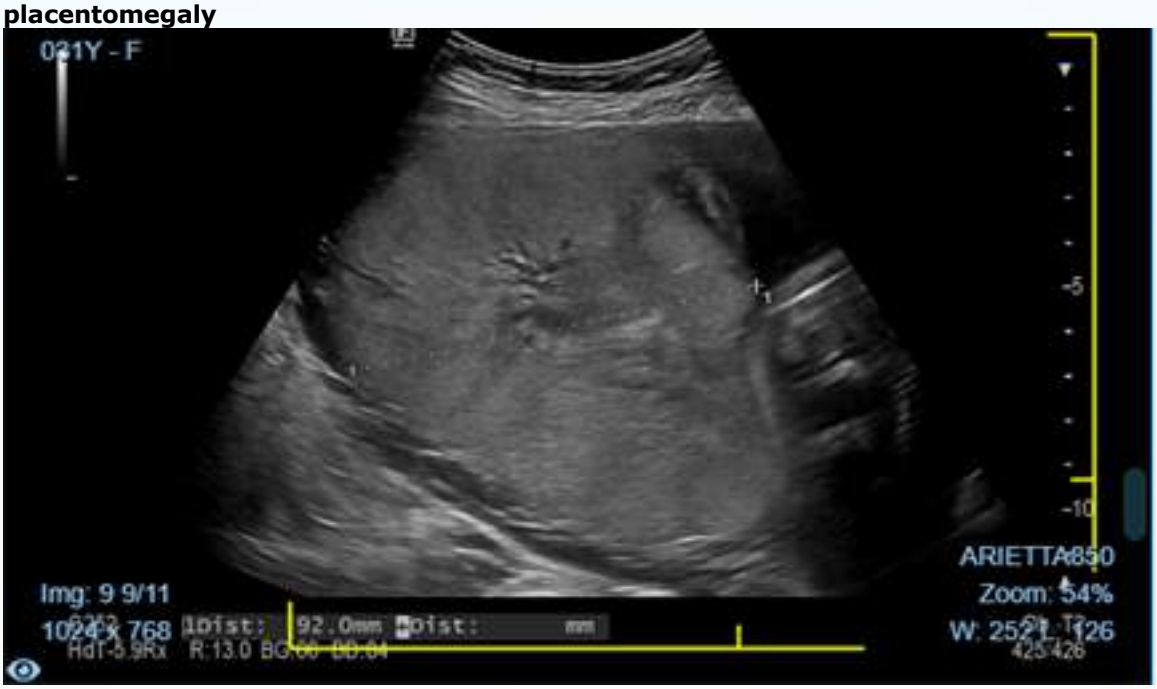
Case Presentation: We report a unique case of prenatal CMT1A diagnosis following intrauterine evaluation for fetal anemia. A 26-week primigravida presented with placentomegaly, single umbilical artery, and severe fetal growth restriction (FGR). Doppler ultrasonography revealed elevated middle cerebral artery peak systolic velocity, prompting cordocentesis, which confirmed fetal anemia and necessitated intrauterine transfusion. Chromosomal microarray analysis identified a 17p12 duplication diagnostic of CMT1A. Subsequent parental testing demonstrated the same duplication in the previously asymptomatic father. The pregnancy was managed to term, resulting in delivery of a growth-restricted but otherwise stable infant. Postnatal genetic testing confirmed CMT1A, and neurodevelopment has remained age-appropriate at 10 months of follow-up.

DISCUSSION: This case is the first to describe prenatal CMT1A diagnosis in association with multiple obstetric complications including fetal anemia, placentomegaly, SUA, and severe FGR. While no established link exists between PMP22 duplication and placental pathology, the case illustrates the importance of comprehensive Doppler surveillance, intrauterine intervention, and advanced genetic testing in complex pregnancies. Furthermore, it highlights the cascade effects of prenatal genetic testing, with the fetus serving as a sentinel for familial diagnosis.

CONCLUSION: Prenatal CMT1A diagnosis remains rare but can emerge through detailed fetal monitoring. This case underscores the value of integrating genetic analysis with perinatal management and cascade family screening.

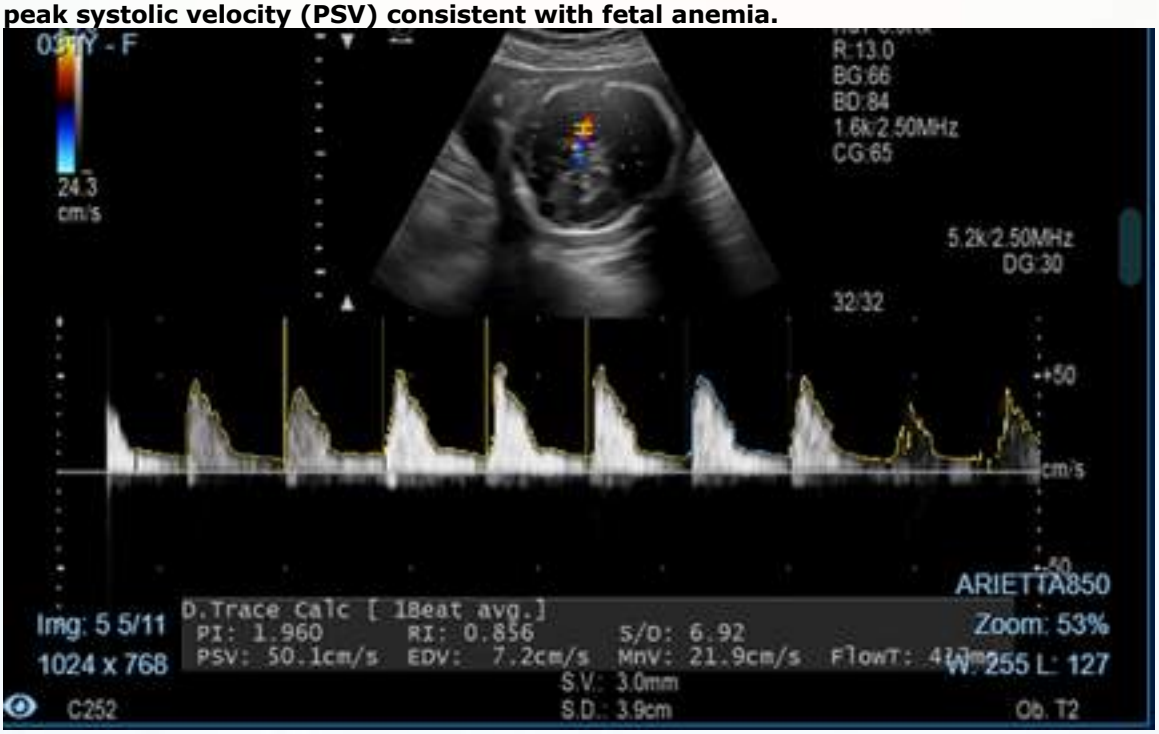
Keywords: Charcot–Marie–Tooth Disease Type 1A, Prenatal Diagnosis, Fetal Anemia

Figure 1: Prenatal ultrasonographic measurement of placental thickness demonstrating placentomegaly



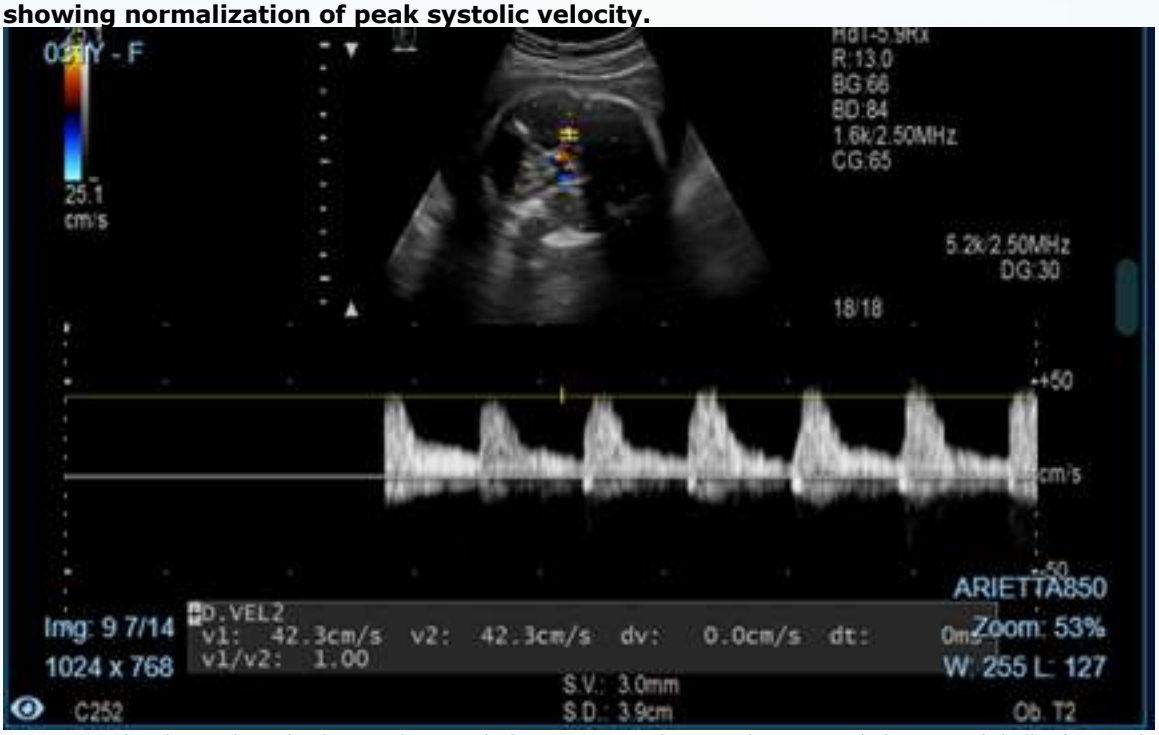
A sagittal view at 26+0 weeks of gestation shows placental thickness of 92.0 mm, which is significantly above the normal range for gestational age

Figure 2: Doppler assessment of the middle cerebral artery (MCA) demonstrating elevated peak systolic velocity (PSV) consistent with fetal anemia.



At 26+1 weeks of gestation, MCA PSV was measured as 50.1 cm/s (1.47 MoM), supporting the diagnosis of moderate fetal anemia prior to intrauterine transfusion.

Figure 3: Post-transfusion Doppler measurement of the middle cerebral artery (MCA) showing normalization of peak systolic velocity.



At 26+1 weeks of gestation, after intrauterine transfusion, MCA PSV decreased to 42.3 cm/s (1.24 MoM), indicating resolution of fetal anemia.

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SS-028

Exit procedure for management of a third trimester fetal cervical teratoma complicated by polyhydramnios: a case report

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[2]Kütahya Şehir Hastanesi, Kadın Hastalıkları ve Doğum Kliniği, Kütahya, Türkiye
[3]Ankara Üniversitesi Tıp Fakültesi Kadın Hastalıkları ve Doğum Kliniği, Perinatoloji Ünitesi, Ankara;Türkiye

OBJECTIVE: Fetal cervical masses are rare but potentially life-threatening anomalies due to airway obstruction at delivery. We present a third-trimester case of a large cervical teratoma managed by the exit procedure.

CASE: A 24-year-old gravida 2 para 1 woman presented at 29+1 weeks with an anterior cervical mass measuring 68×68 mm. At 31+2 weeks, the mass enlarged to 78×91 mm with arterial flow (PI 3.1, PSV 83.3), and polyhydramnios was noted. At 34+4 weeks, polyhydramnios progressed (AFI 210 mm) and the mass increased to 106×78 mm. Due to worsening contractions, the exit procedure was performed. A 3200 g neonate was delivered via cesarean section, successfully intubated under placental support, and underwent mass excision. Histopathology confirmed a mature teratoma. The neonate required intensive care after surgery. Genetic testing was not performed.

CONCLUSION: Fetal cervical teratomas may enlarge rapidly and be complicated by polyhydramnios. Even when diagnosed in the late third trimester, ultrasound follow-up and multidisciplinary planning are crucial. The exit procedure provides a safe perinatal strategy to secure the airway and allow resection, improving survival. This case illustrates that giant cervical teratomas diagnosed late in pregnancy can still be managed successfully with prenatal surveillance and coordinated perinatal care. The report emphasizes referral to tertiary centers where maternal–fetal medicine, neonatology, anesthesiology, and pediatric surgery collaborate. Awareness of this rare condition may increase preparedness and improve outcomes. Additional data and follow-up are needed to clarify prognosis, refine surgical approaches, and assess the role of prenatal genetic testing.

Anahtar Kelimeler: cervical teratoma, exit procedure, fetal tumor, polyhydramnios, prenatal diagnosis, ultrasound

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SS-029

Ellis-van Creveld Syndrome: A Case Report

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[2]Perinatology Clinic, Adana City Hospital, Adana, Turkey

INTRODUCTION: Ellis-van Creveld syndrome (EvC) is a rare autosomal recessive ciliopathy characterized by short ribs and limbs, chondroectodermal dysplasia, postaxial polydactyly, and congenital heart defects in nearly 60% of cases (OMIM 225500). We report a case in which typical sonographic findings raised suspicion for skeletal dysplasia and cardiac anomaly, but definitive diagnosis was delayed until after birth due to parental refusal of invasive testing.

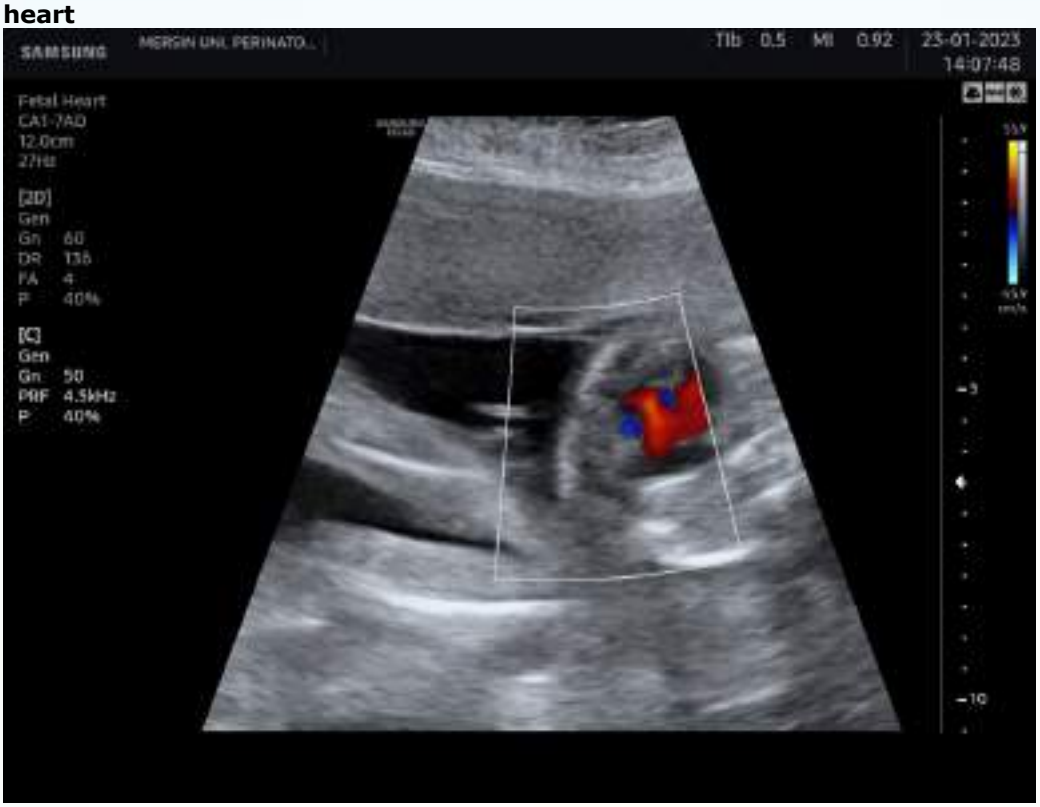
Case Presentation: A 35-year-old gravida 5, para 4 woman was referred following increased nuchal translucency detected in the first trimester. Detailed ultrasound revealed nasal bone hypoplasia (0.1p), shortening of femur and humerus (<5p), femoral bowing, narrow thorax with elevated cardiothoracic ratio, muscular ventricular septal defect (VSD), and postaxial polydactyly. Invasive diagnostic testing was proposed but declined.

At 35+6 weeks of gestation, cesarean delivery was performed for severe preeclampsia, polyhydramnios, and breech presentation. Postnatal evaluation confirmed polydactyly, syndactyly, short-limb dwarfism, narrow thorax, and muscular VSD. Karyotype analysis was normal. Whole-exome sequencing identified a homozygous pathogenic variant in the EVC gene (NM-153717.3), confirming Ellis-van Creveld syndrome within the spectrum of Weyers acrofacial dysostosis. The newborn required prolonged neonatal intensive care due to asphyxiating thoracic dystrophy. At 28 months of age, the child remains under follow-up for severe growth retardation, cardiac complications, and orthopedic problems.

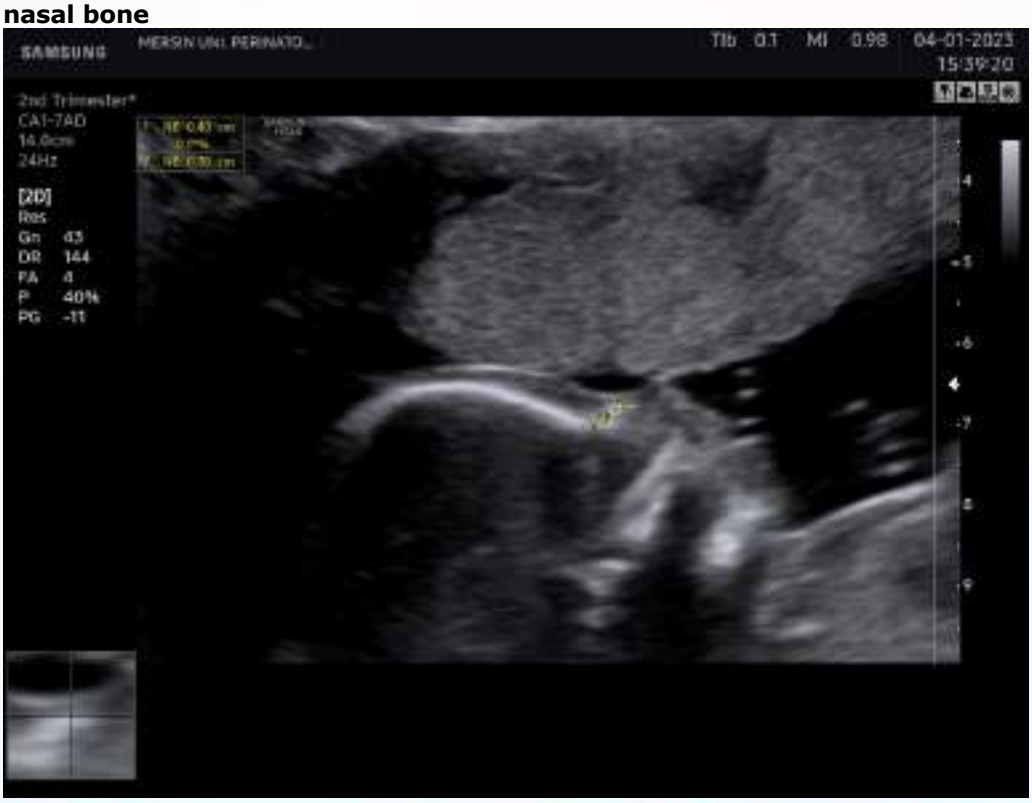
CONCLUSION: EvC syndrome is extremely rare (1 in 1,000,000 live births) and should be suspected when characteristic prenatal findings are present. Even in the context of a normal karyotype, whole-exome sequencing provides a definitive diagnosis and is valuable for prognostication and genetic counseling.

Keywords: Ellis-van Creveld syndrome, whole-exome sequencing, skeletal dysplasia, polydactyly, ventricular septal defect

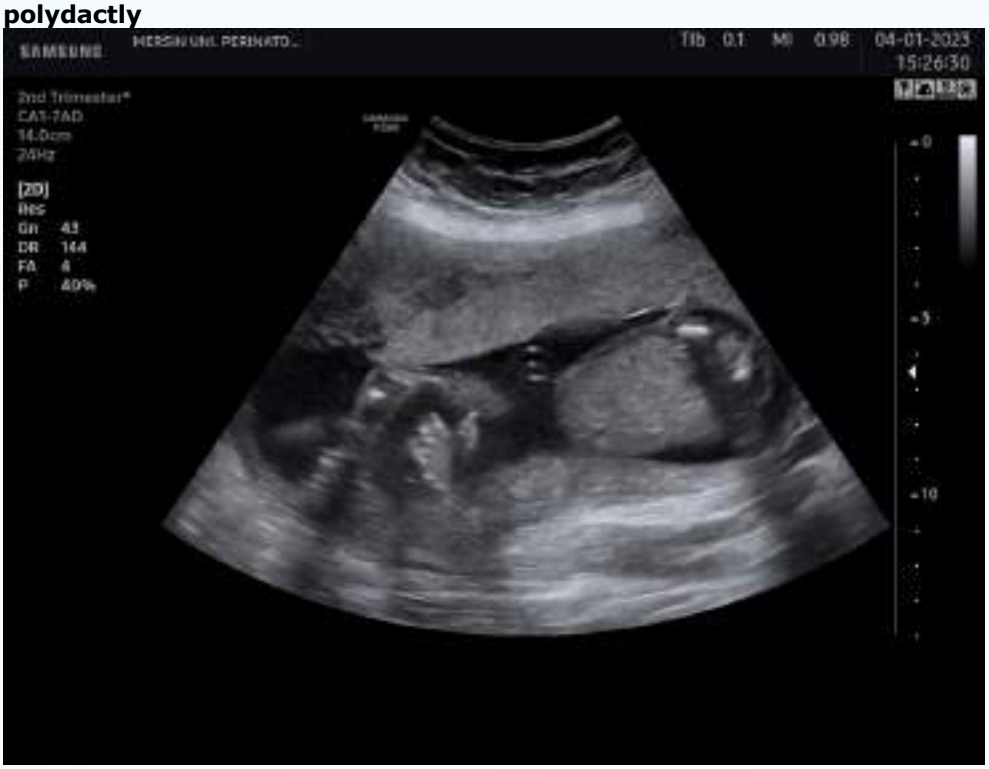
heart



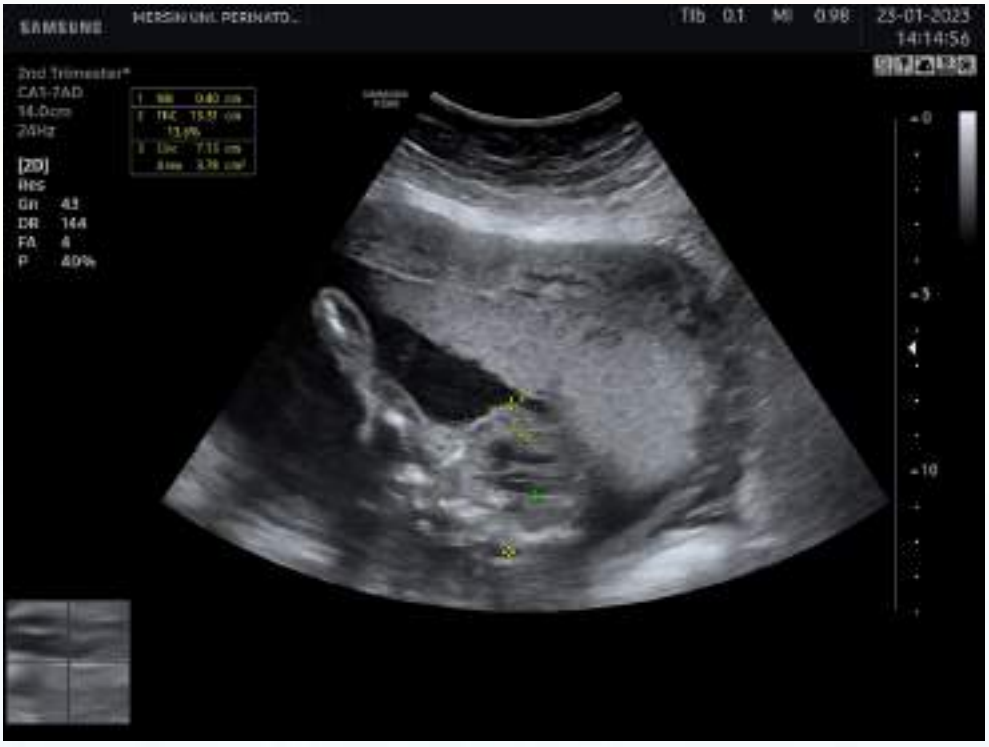
nasal bone



polydactly



thorax



SS-030

Prenatal Detection of Ectrodactyly-Ectodermal Dysplasia-Clefting Syndrome with Multiple Congenital Anomalies in a Consanguineous Pregnancy

Reşat Mısırlıoğlu, Filiz Yarsilikal Guleroglu, Ali Cetin
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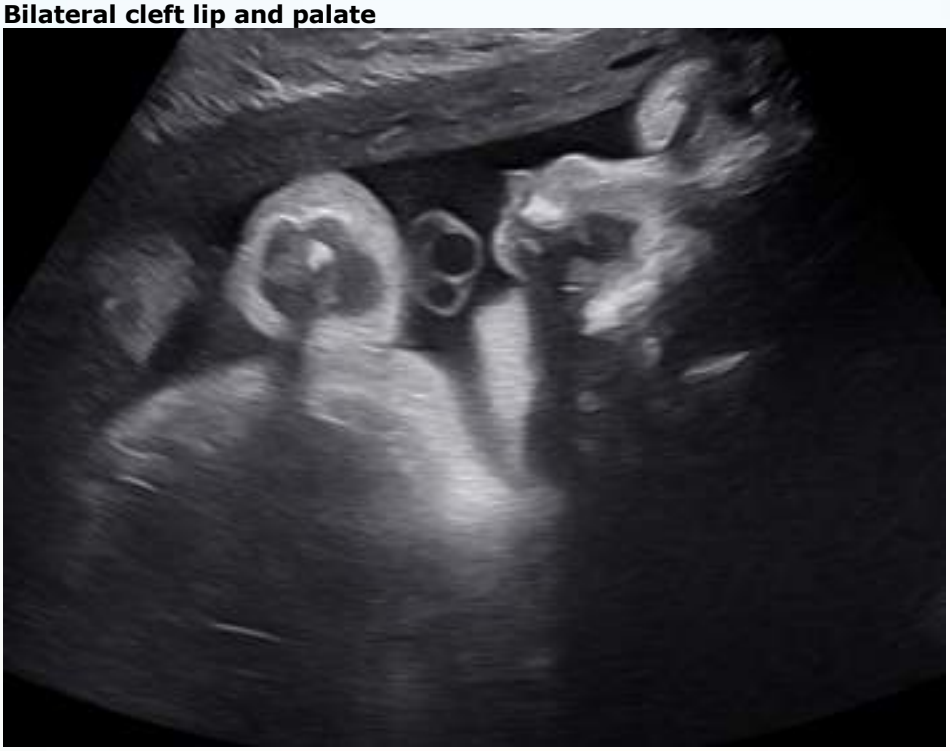
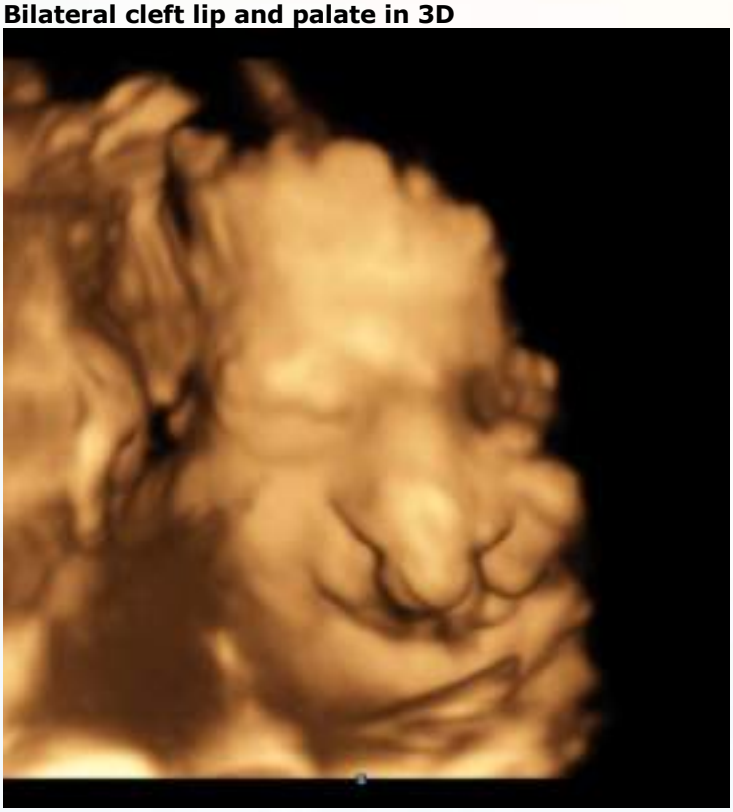
INTRODUCTION: Ectrodactyly-ectodermal dysplasia-clefting (EEC) syndrome is a rare autosomal dominant disorder, primarily caused by mutations in TP63. It is characterized by variable expression of split-hand/foot malformations, craniofacial clefts, and ectodermal dysplasia, with occasional cardiac and genitourinary anomalies. Prenatal recognition is essential for counseling and management. CASE: We present a 21-year-old primigravida with a consanguineous marriage (husband is her cousin), who had not undergone first- or second-trimester screening. Based on the last menstrual period (January 28, 2025), the fetus demonstrated intrauterine growth restriction (abdominal circumference <1st percentile) with normal amniotic fluid. Detailed ultrasonography revealed hypoplastic nasal bone (<1st percentile), bilateral cleft lip and palate, ectrodactyly of the right hand and foot, and two ventricular septal defects (muscular and apical). These findings were highly suggestive of EEC syndrome. No additional systemic anomalies were observed. Genetic testing was declined by the family. DISCUSSION: EEC syndrome demonstrates marked phenotypic variability, even within families, and can be sporadic or inherited. Prenatal ultrasound plays a pivotal role in raising suspicion, particularly when multiple features such as clefting, limb anomalies, and cardiac defects coexist. While TP63 mutations confirm the diagnosis, sonographic pattern recognition remains crucial in resource-limited settings. Early identification facilitates parental counseling, perinatal planning, and genetic evaluation. CONCLUSION: This case underscores the importance of detailed mid-trimester imaging in detecting rare syndromic associations. Multidisciplinary evaluation and genetic counseling are essential for optimal management and recurrence risk assessment.

Keywords: ectrodactyly, cleft lip and palate, prenatal diagnosis,

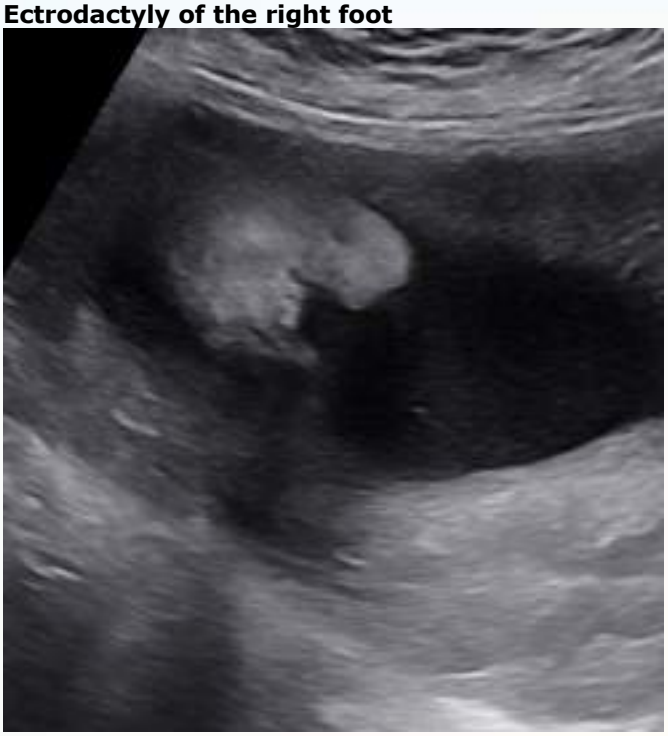
Bilateral cleft lip and palate



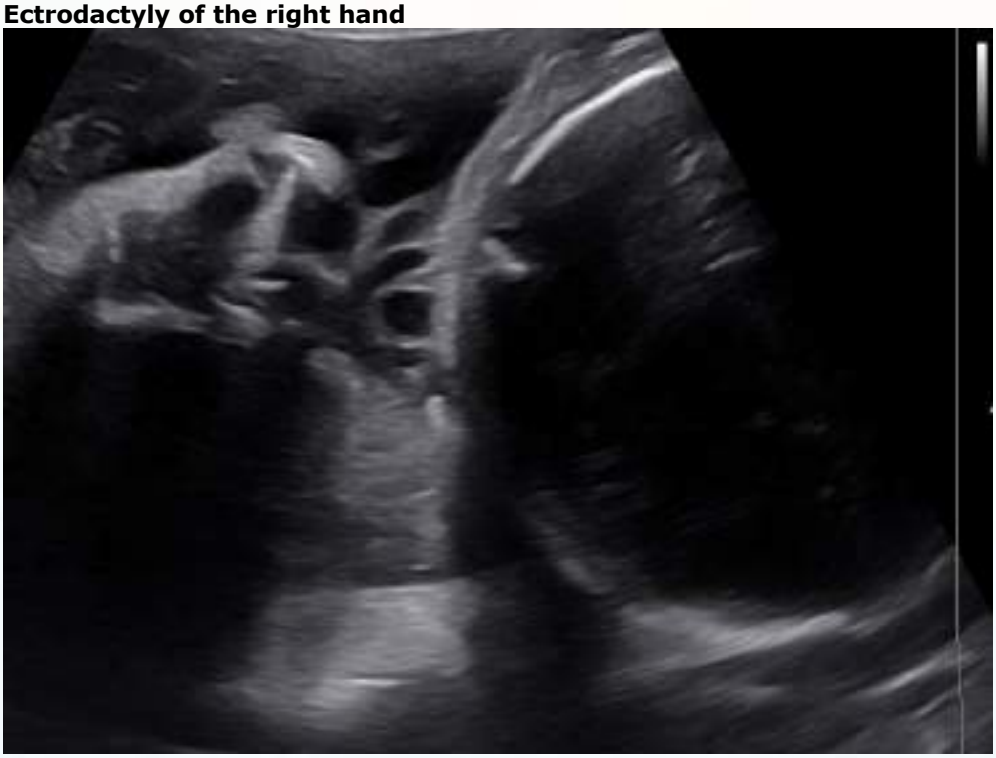
Bilateral cleft lip and palate in 3D



Ectrodactyly of the right foot



Ectrodactyly of the right hand



SS-031

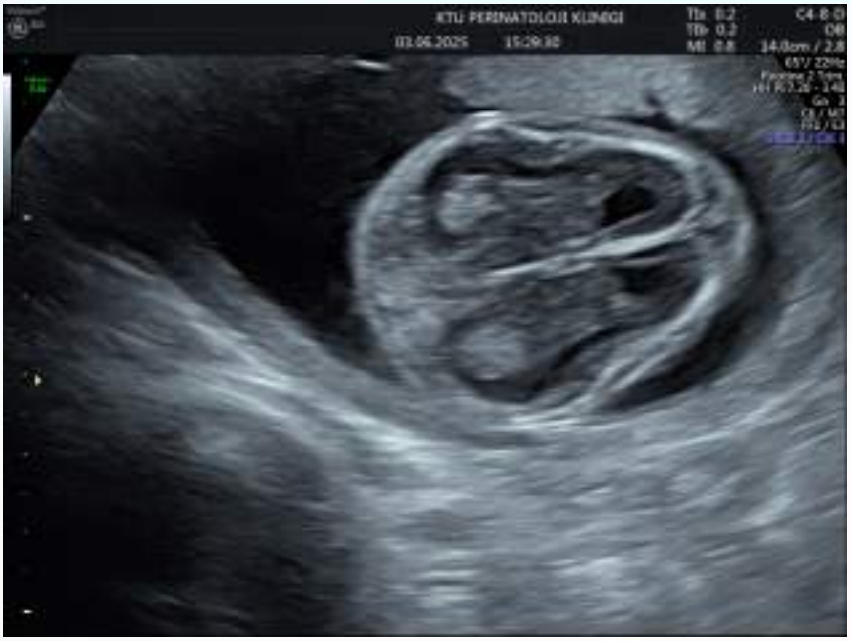
Lethal Osteogenesis Imperfecta Type II

Mensure Tonguc, Emine seda Guvendag guven, Ayberk turkyilmaz
Karadeniz Teknik Üniversitesi Kadın Hastalıkları ve Doğum Ana Bilim Dalı Perinatoloji Bölümü Trabzon

This report details a case of lethal Osteogenesis Imperfecta (OI) Type II, diagnosed early in the second trimester, emphasizing the integrated approach of prenatal imaging, systematic differential diagnosis, and comprehensive genetic analysis. The patient, a 32-year-old gravida 4, para 1, abortus 2, was referred for a second-trimester obstetric ultrasound, which revealed severe fetal skeletal anomalies. Key findings included marked micromelia, severe hypomineralization of the skull, a hypoplastic thorax, and bowing of the long bones. Based on this constellation of findings, a differential diagnosis of lethal skeletal dysplasias was established. Amniocentesis and subsequent genetic testing, including Whole Exome Sequencing (WES), were performed to confirm the diagnosis and to exclude other similar conditions. Analysis ruled out Thanatophoric dysplasia and other differential diagnoses. The definitive diagnosis was confirmed by the identification of a heterozygous pathogenic variant in the COL1A1 gene, specifically a c.2228GGT (p.g743v) mutation. Following extensive genetic counseling, the family opted for a termination of the pregnancy at 23 weeks and 3 days of gestation. Post-mortem examination corroborated the prenatal findings, highlighting a narrow thorax, generalized micromelia, and characteristic skeletal deformities. This case underscores the critical importance of a multi-faceted diagnostic strategy for severe prenatal skeletal disorders and illustrates the clinical significance of a specific glycine substitution in the collagen protein structure. It also highlights the value of comprehensive genetic testing in identifying incidental findings with important implications for future genetic counseling.

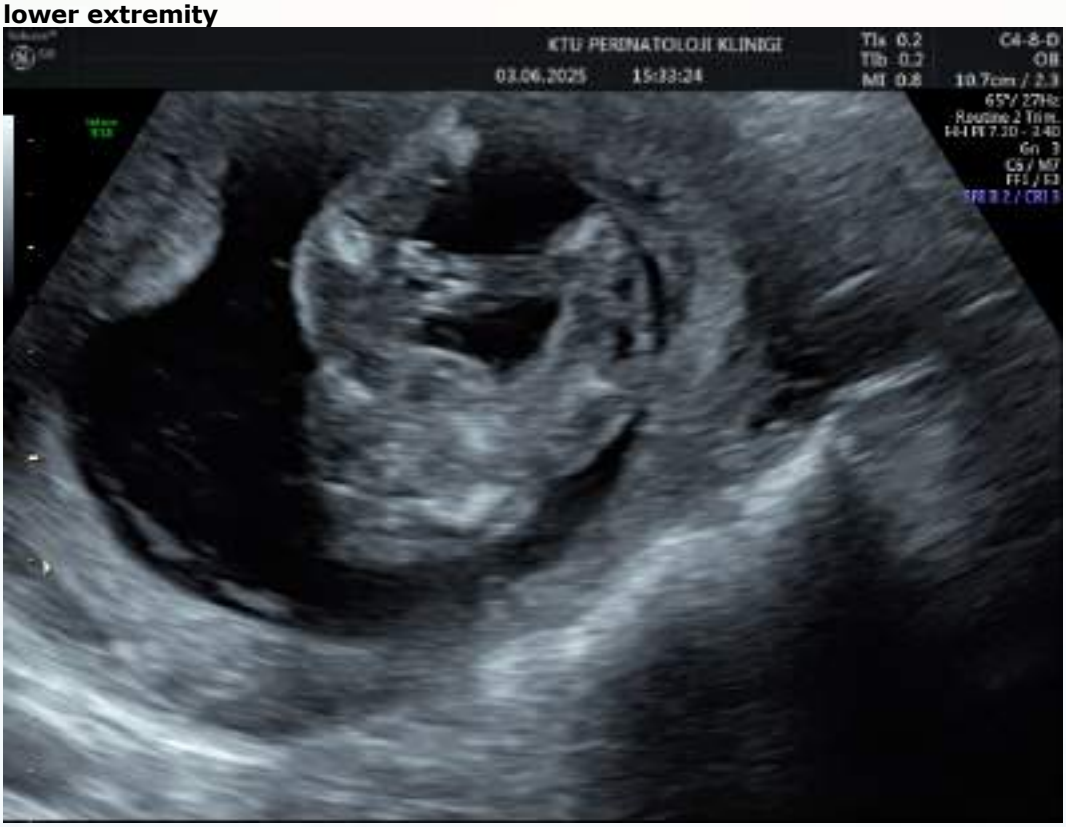
Keywords: Osteogenesis Imperfecta (OI) Type II,COL1A1 gene mutation, skeletal dysplasia

fetal head



Lethal Osteogenesis Imperfecta (OI)

lower extremity

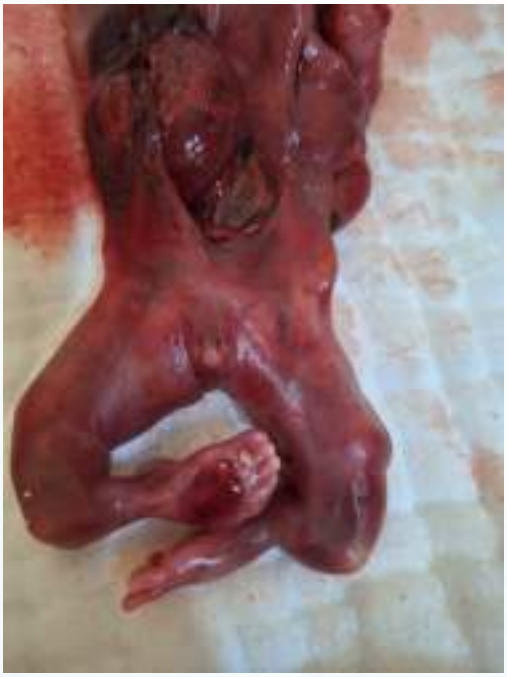


Lethal Osteogenesis Imperfecta (OI)

postmortem face

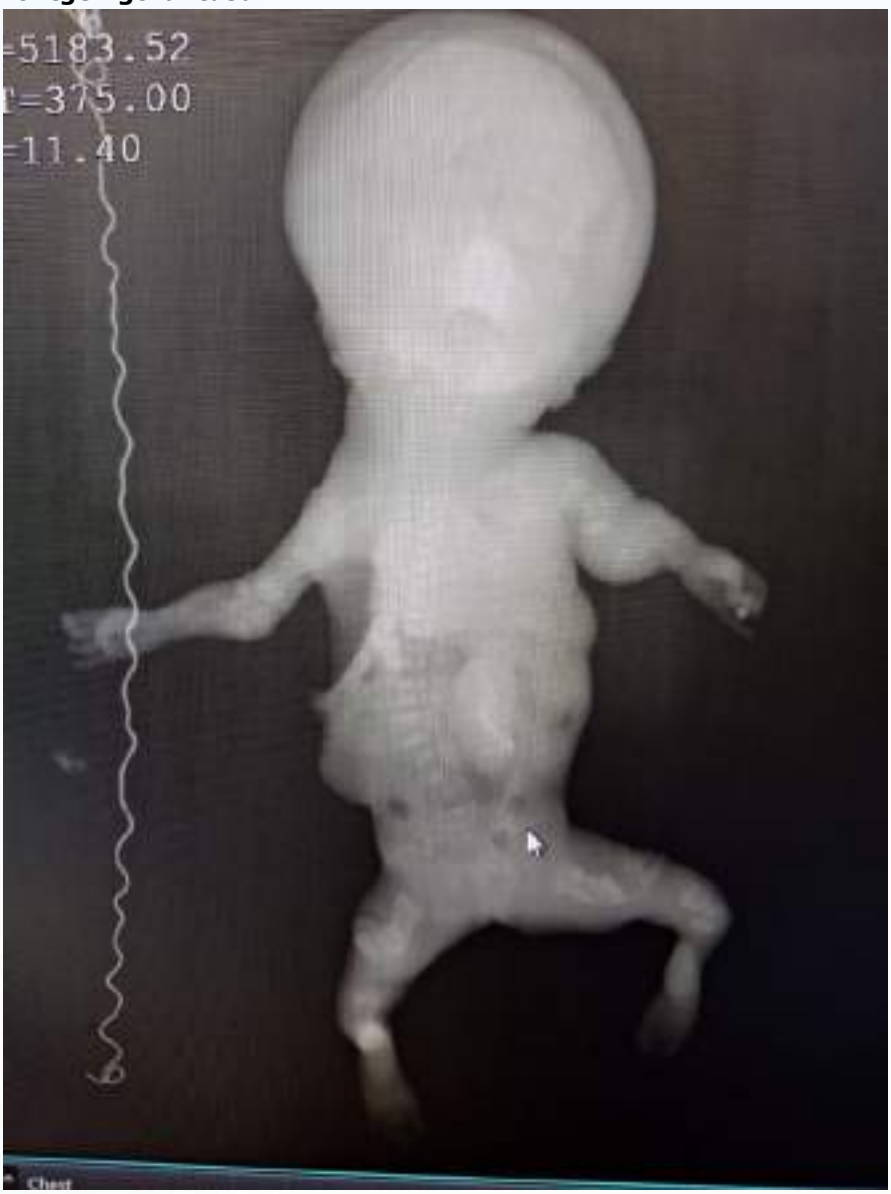
Lethal Osteogenesis Imperfecta (OI)

Postmortem Lowe extremity



Lethal Osteogenesis Imperfecta (OI)

röntgen görüntüsü



Lethal Osteogenesis Imperfecta (OI)

SS-032

Nadir bir toraks hipoplazisi nedeni olarak Raine Sendromu

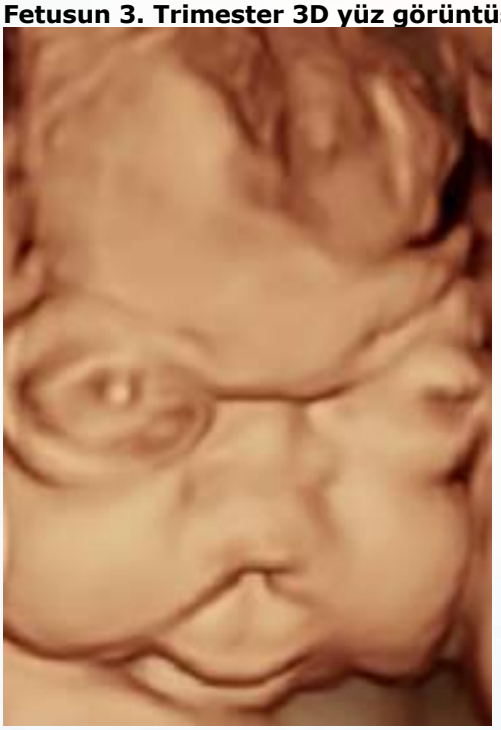
Coşkun Ümit
Ankara Atatürk Sanatoryum Eğitim ve Araştırma Hastanesi, Ankara

23 yaşında G2P1 hasta 27. Haftada tespit edilen uzun kemik kısalığı ve ekzoftalmus nedeniyle perinatoloji kliniğine başvurdu. Yapılan ultrasonda hipertelorizm, mikrognati, koanal atrezi, midface hipoplazi, ekzoftalmus, mikrosefali, intrakranial yaygın kalsifikasyonlar, toraks hipoplazisi, uzun kemik kısalığı tespit edildi. Klinik bulgular Raine Sendromu ile uyumluydu. Yapılan amniosentezde FAM20C geninde c.708C>A (p.Tyr236Ter) varyantı homozigot olarak tespit edildi. Hastaya olumsuz prognoz hakkında bilgi verildi ve gebelik terminasyonu önerildi. Hasta terminasyonu kabul etmedi, 38. Haftada 1 adet canlı, 2000 gram erkek bebek doğurtuldu.

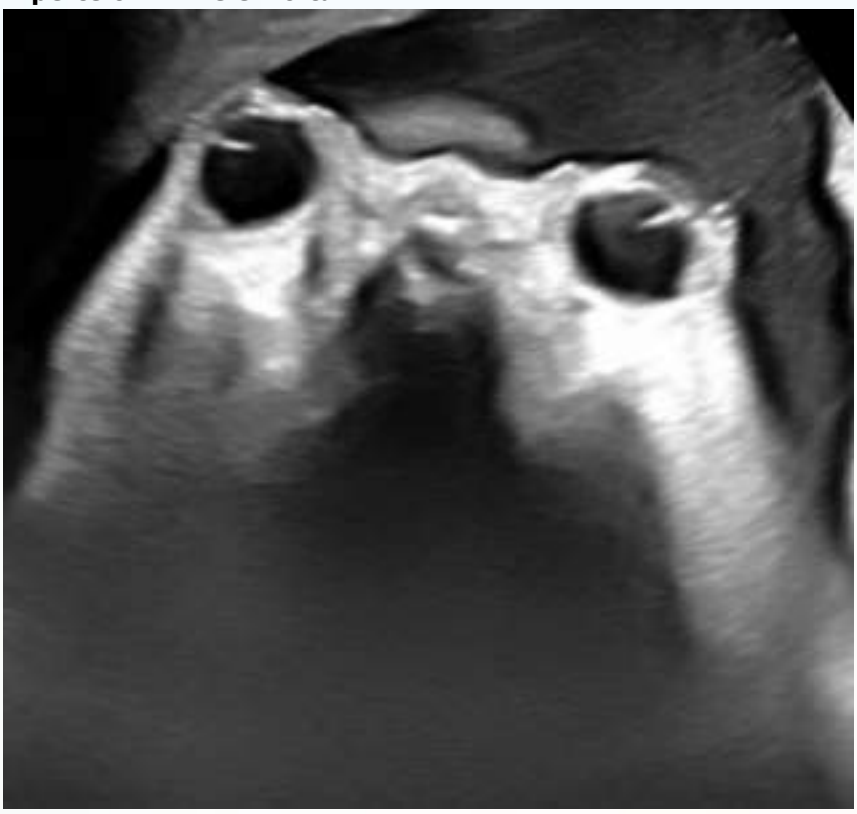
Raine sendromu, ekzoftalmi, koanal atrezi veya stenoz, osteoskleroz ve serebral kalsifikasyonlar gibi karakteristik özellikleri olan çok nadir görülen bir genetik hastalıktır. Sıklığı yaklaşık olarak 1/1000000 dur. Bu olgu nadir görülen Raine sendromunun prenatal tanısı, ultrasonografik bulguları, takibi ve yönetimi açısından önemli bir örnek teşkil etmektedir.

Anahtar Kelimeler: Ekzoftalmi, fetus, genetik, prenatal tanı, Raine, ultrason

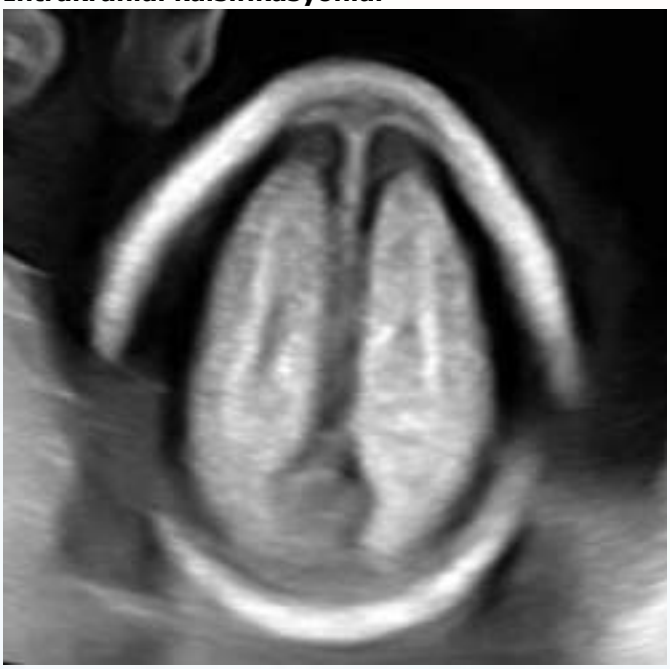
Fetusun 3. Trimester 3D yüz görüntüsü



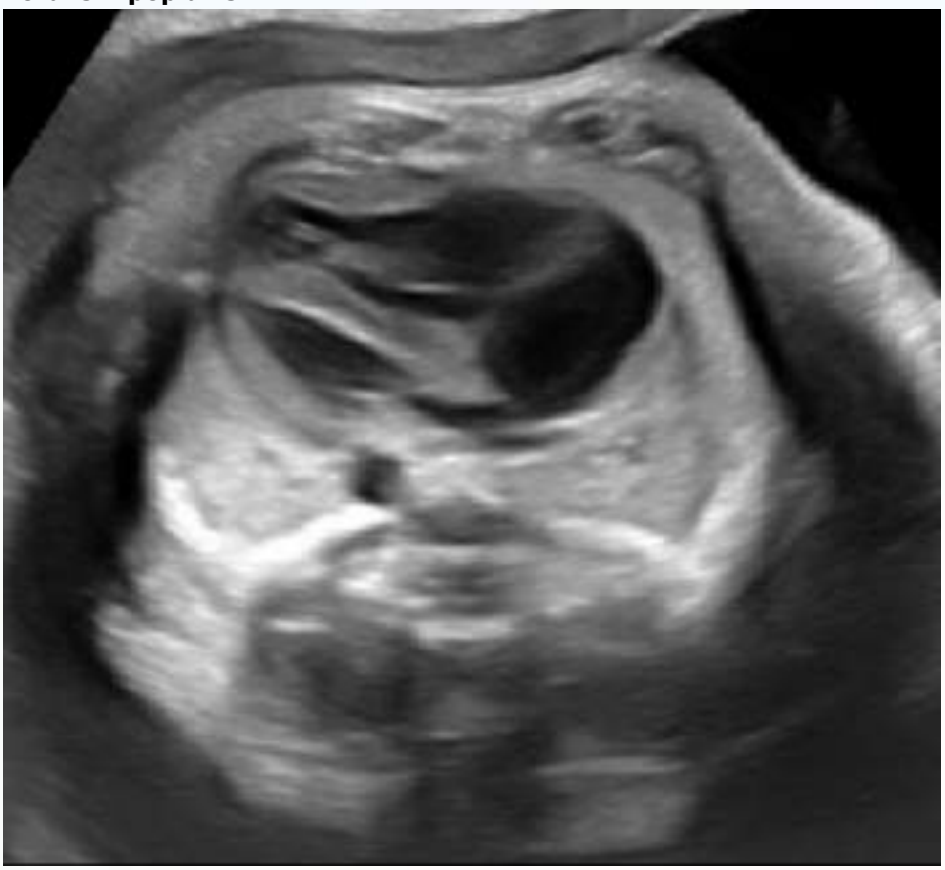
Hipertelorizm ve ekzoftalmi



İntrakranial kalsifikasyonlar



Toraks hipoplazisi



SS-033

Kalıcı fetal bradikardi için salbutamol kullanımı olgu sunumu

Sevinç Rabia Serindağ, Erkan Çağlıyan
Dokuz Eylül Üniversitesi Tıp Fakültesi, Perinatoloji Anabilim Dalı, İzmir

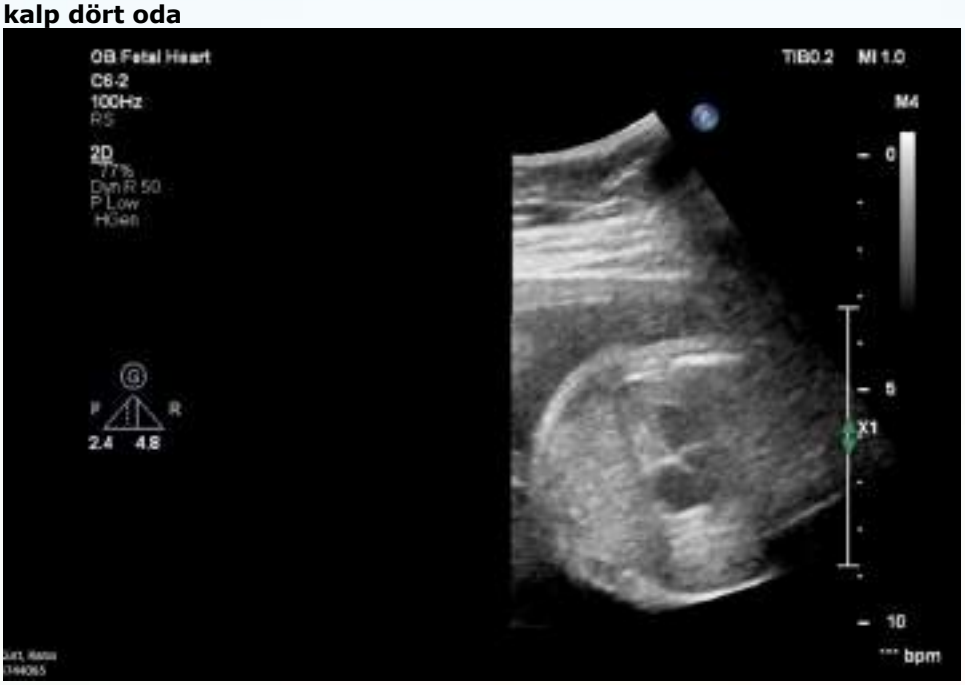
AMAÇ: İzole dekstrokardi, kalp, göğüste normalden daha sağda bulunduğu bir situs anomalisidir. Genellikle ciddi kalp defektleri ve pulmoner hipoplazi gibi kalp ile ilgili anormallikler ile ilişkilidir. Olgumuzda izole dekstrokardisi oaln kliniğimize 23. Gebelik haftasında sağ atrial izomerizm ve hidrops fetalis olmaksızın fetal bradiklardi gelişen fetüsü sunmayı planladık.

OLGU: G:1 p:0 olan gebe 22. Gebelik haftasında kliniğimize sağ atrial izomerizim tanısı ile dış merkezden invazif tanı testi yapılması ve değerlendirilmesi için yönlendirildi. Hastanın yapılan prenatal testlerinde herhangibir anomali izlenmedi. Birinci trimester yapılan kombine tarama testinde düşük risk olarak değerlendirildi. Hasta 20. gebelik haftasında fetal anomali taramasında kalbin apeksinin sağda izlenmesi üzerine kliniğimize yönlendirildi Hastaya gebelik haftası göz öninde bulundurularaka amniosentez ile karyotip analizi yapıldı. Kromozom sayısı 46 ile uyumlu bulundu merkezimizde mikroARRAY yapılmadığı bilgisi verildi. Hasta Amniyosentez sonucu ile perinatoloji konseyinde değerlendirildi.Çocuck kardiyolojisine fetak EKO için yönlendirildi. Dekstrokardi, biventriküler hipertrofi ve bradikardi doğrulandı; Çocuck kardiyolojisinin önerisi ile sallbutamol başlandı. Hasta 39. Gebelik haftasında membran rüptürü ve makat geliş nedeniyle sezaryen ile doğurtuldu. Apgar 6-82400 gr kız bebek doğurtup pyenidoğan ekiplerine teslim edildi.

SONUÇ: Fetal dekstrokardi perinatal mortalitesi yüksek bir prenatal ultrasonografi bulgusudur. Çoğunlukla kompleks kardiyak anomaliler ile birliktedir. Fetal dektrokardili fetüslerin çocuk kardiyoloğu ve perinatolog tarafından ekip anlayışı içinde takipleri uygun olacaktır.

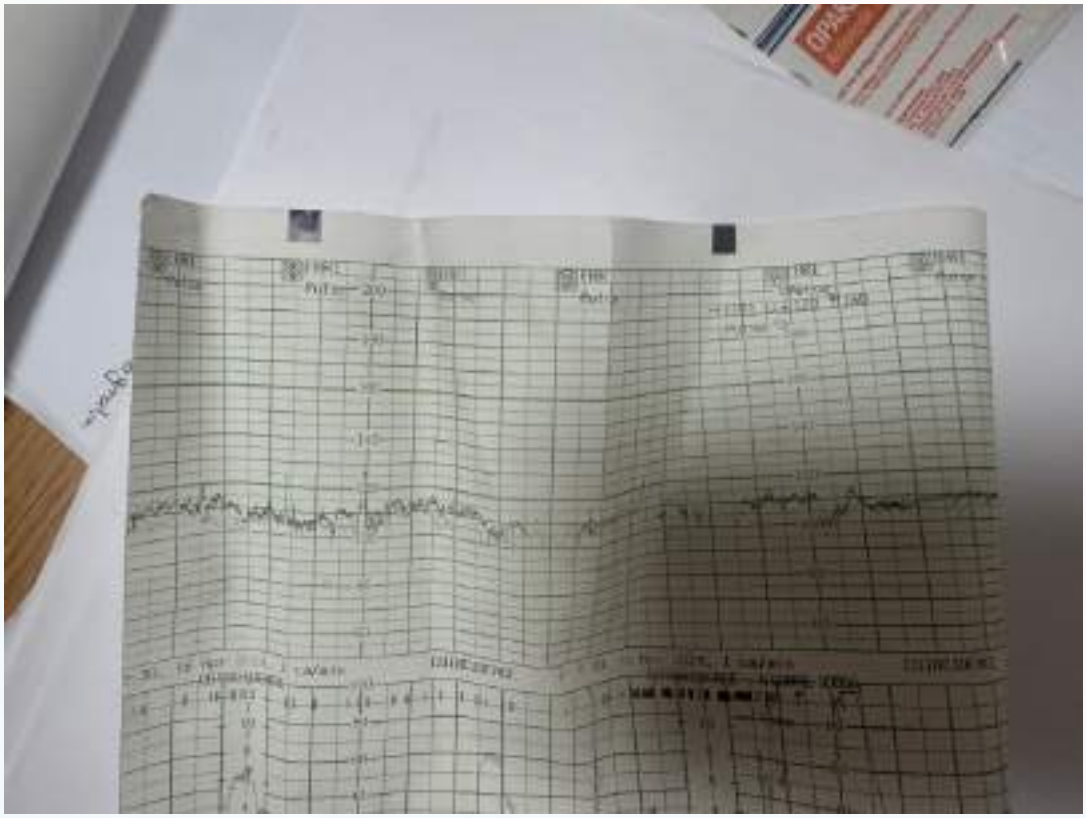
Anahtar Kelimeler: Dextrokardi, sağ atriyal izomerizm, fetal bradikardi;

kalp dört oda



dört oda görüntüsünde dekstrokardi ve biventriküler hipertrofi

nst



bradikardi

Türkiye Maternal Fetal Tıp ve Perinatoloji Derneği
Ultrasonografi Kongresi
29 Ekim - 1 Kasım 2025
Renaissance Polat İstanbul Hotel - Yeşilköy

SS-034

Assessment Of Early And Late Neonatal Outcomes Associated With Antenatal Magnesium Sulfate Neuroprotection At Varying Durations İn Preterm Births Before 32 Weeks Gestation

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[2]Sakarya Eğitim ve Araştırma Hastanesi Kadın Hastalıkları ve Doğum Anabilim Dalı, Perinatoloji Bilim Dalı, Sakarya
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Introduction and AIM: Early preterm birth increases the risk of neurodevelopmental problems. Magnesium sulfate given to mothers before delivery may offer neuroprotection, even if the full 24-hour dose isn't completed. This study aimed to assess early and late neurodevelopmental outcomes in preterm infants exposed to antenatal $MgSO_4$.

MATERIALS-METHODS: Between January 2019 and December 2022, infants born before 32 weeks of gestation at Sakarya University Training and Research Hospital were screened. Only infants who received antenatal $MgSO_4$ were included. The study group was divided into three subgroups: those who received the standard 24-hour dose, who received it for a shorter duration, and delivered after a certain interval post-treatment. A control group of infants who did not receive $MgSO_4$ was also included. The study assessed the duration of $MgSO_4$ administration, gestational age and serum magnesium levels. Late neurodevelopmental outcomes were measured using the Denver-2 test.

RESULTS: While gestational age and maternal factors were similar across groups, significant differences were found in NICU stay, APGAR scores, serum magnesium, and Denver-2 outcomes. Cerebral palsy was observed only in the control group.

Neonatal outcomes varied depending on the duration of antenatal $MgSO_4$ administration, indicating its influence on treatment effectiveness.

CONCLUSION: In preterm infants whose mothers received antenatal $MgSO_4$, significant differences were observed in both early and late developmental outcomes, including APGAR scores, NICU stay, early sepsis, NEC, ROP and cerebral palsy with differences related to the duration of MgSO4 administration. This indicates that the duration of MgSO4 administration affects the effectiveness of the treatment.

Anahtar Kelimeler: Preterm, MgSO4, Denver-2

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Ultrasonografi Kongresi
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SS-035

"Association of birth weight with neonatal intensive care admissions: A retrospective analysis of 1256 deliveries"

Mustafa Koçar[1], Onur Burak Cegilli[2], Engin Öztürk[3], Gulsah Aynaoğlu Yıldız[3]
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[2]Simav Doç. Dr. İsmail Karakuyu Devlet Hastanesi, Simav, Kütahya, Türkiye
[3]Ankara Üniversitesi Tıp Fakültesi Kadın Hastalıkları ve Doğum Kliniği, Perinatoloji Ünitesi, Ankara;Türkiye

OBJECTIVE: Neonatal intensive care unit (NICU) admissions vary by birth weight and timing. This study evaluated their distribution and primary diagnoses according to birth weight groups and early (≤2 days) versus late (>2 days) admission.

METHODS: A retrospective review included 1256 singleton deliveries at Simav Hospital (2015–2019). Neonates were grouped as <2500 g, 2500–3999 g, and ≥4000 g. NICU admissions were classified as early (0–2 days) or late (>2 days). Diagnostic indications were obtained from records, and in cases with multiple diagnoses, the primary indication was selected. Routine child health examination entries were excluded.

RESULTS: Among neonates <2500 g, 68.6% required NICU admission, mostly early (79%) due to respiratory distress, transient tachypnea of the newborn (TTN), and hypoglycemia. In the 2500–3999 g group, 39.3% were admitted, with 38% late, mainly for hyperbilirubinemia and feeding problems. For neonates ≥4000 g, 33.3% needed NICU care, of which 82% were early, primarily for TTN and hypoglycemia. Early admissions were dominated by respiratory and metabolic complications, whereas late admissions were largely due to jaundice, feeding problems, and sepsis. NICU admissions were highest in low birth weight infants and lower in normal and high weight groups.

CONCLUSION: NICU admissions were strongly associated with birth weight. Low birth weight infants had the highest risk, while normal and macrosomic infants showed reduced rates. Early admissions were mainly linked to respiratory and metabolic issues, whereas late admissions were driven by jaundice and infections. These findings emphasize the need for monitoring vulnerable neonates, especially those with low birth weight.

Anahtar Kelimeler: birth weight, hyperbilirubinemia, hypoglycemia, neonatal intensive care unit, respiratory distress, retrospective study

Figure 1. NICU admission rates by birth weight group

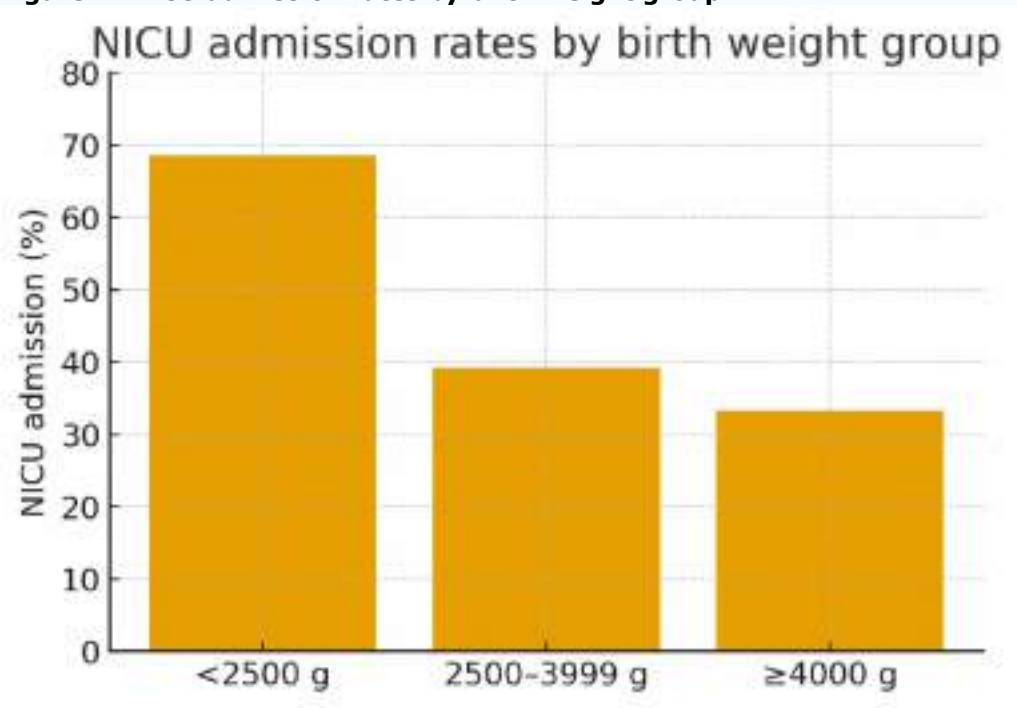

Figure 2. Timing of NICU admission by birth weight group

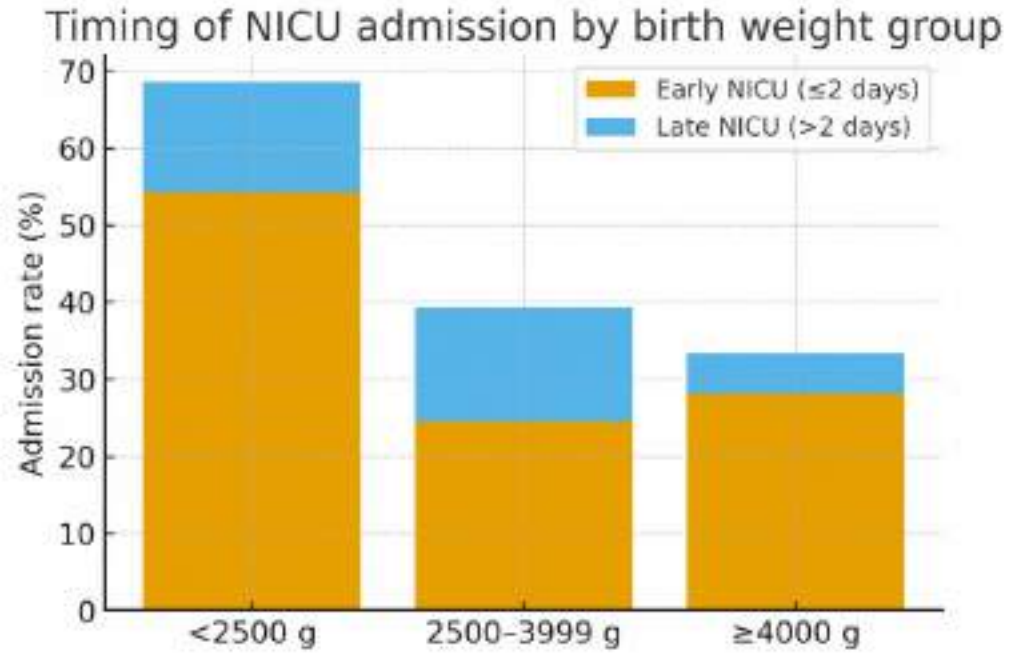

Figure 3. Primary diagnoses in early NICU admissions (≤2 days, N=321)

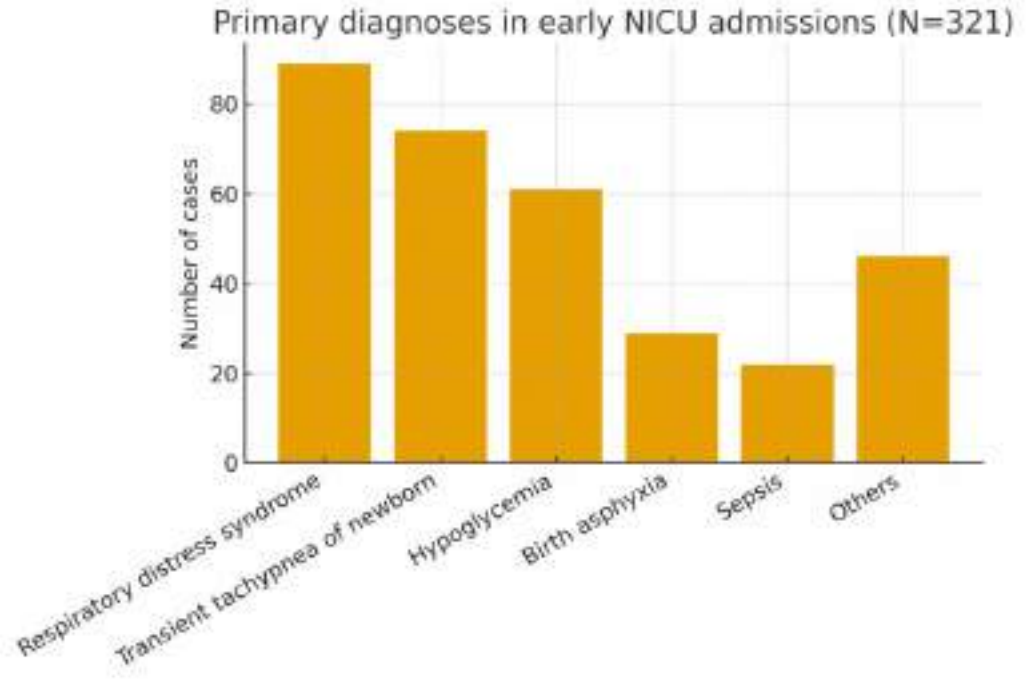

Figure 4. Primary diagnoses in late NICU admissions (>2 days, N=179)

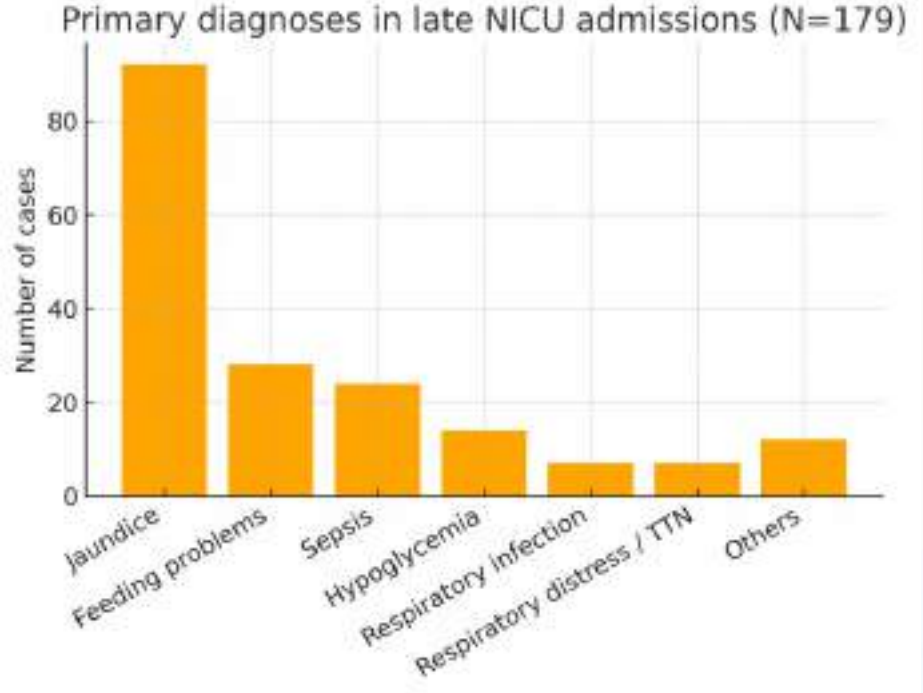

Figure 5 – Birth weight and NICU admission pattern

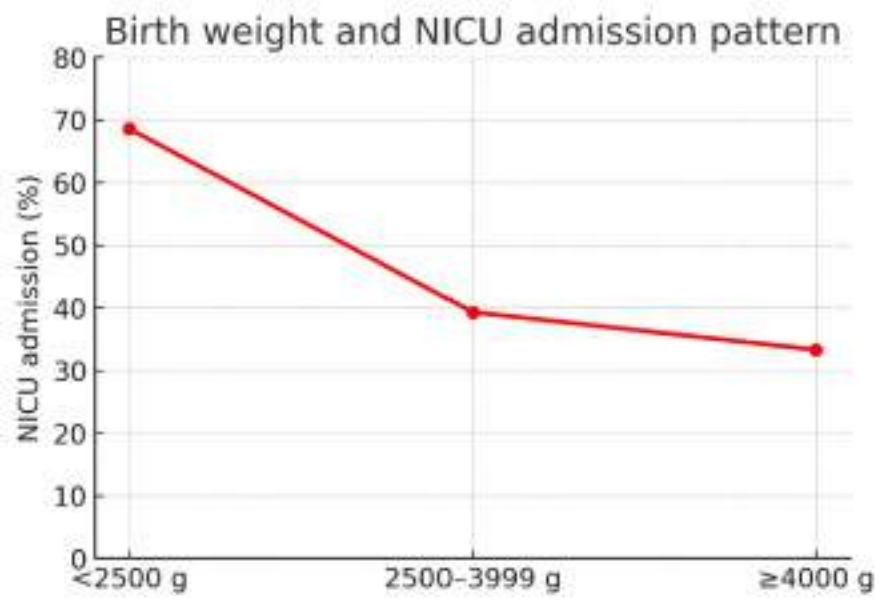

Table 1. Baseline characteristics of the study population (N=1256)

Variable	Value
Maternal age (years), mean ± SD	28.6 ± 5.2 (17–45)
Gestational age (weeks), mean ± SD	38.9 ± 1.4 (24–42)
Birth weight (g), mean ± SD	3290 ± 427 (680–4740)
Birth weight <2500 g	35 (2.8%)
Birth weight 2500–3999 g	1164 (92.7%)
Birth weight ≥4000 g	57 (4.5%)
Total NICU admissions	500 (39.8%)
Early NICU (≤2 days)	321 (25.6%)
321 (25.6%)	179 (14.3%)
No NICU admission	756 (60.2%

Table 2. NICU admission status according to birth weight groups

Birth weight group	Early NICU (≤2 days)	Late NICU (>2 days)	No admission	Total
<2500 g (N=35)	19 (54.3%)	5 (14.3%)	11 (31.4%)	35
2500–3999 g (N=1164)	286 (24.6%)	171 (14.7%)	707 (60.7%)	1164
≥4000 g (N=57	16 (28.1%)	3 (5.3%)	38 (66.7%)	57
Total (N=1256	321 (25.6%)	179 (14.3%)	756 (60.2%)	1256

TTN and hypoglycemia. kelimesinden sonra

able 4. Primary diagnoses in early NICU admissions according to birth weight groups

Diagnosis	<2500 g (N=35)	2500–3999 g (N=1164)	≥4000 g (N=57)	Total (N=321)
Respiratory distress syndrome	18 (51.4%)	62 (21.7%)	9 (56.3%)	89 (27.7%)
Transient tachypnea of newborn	7 (20.0%)	61 (21.3%)	6 (37.5%)	74 (23.1%)
Hypoglycemia	3 (8.6%)	44 (15.3%)	14 (87.5%)	61 (19.0%)
Birth asphyxia	4 (11.4%)	23 (8.0%)	2 (12.5%)	29 (9.0%)
Sepsis	2 (5.7%)	18 (6.3%)	2 (12.5%)	22 (6.9%)
Others	1 (2.9%)	77 (27.4%)	0 (0%)	46 (14.3%)
Total	35 (100%)	285 (100%)	33 (100%)	321 (100%)

hypoglycemia and TTN as admission causes. cümlesinden sonra

Table 3. Most frequent primary diagnoses in early and late NICU admissions

Diagnosis	Early NICU (≤2 days)	Late NICU (>2 days)
Respiratory distress	112 (34.9%)	12 (6.7%)
Transient tachypnea of newborn	85 (26.5%)	9 (5.0%)
Hypoglycemia	67 (20.9%)	18 (10.1%)
Asphyxia	34 (10.6%)	2 (1.1%)
Sepsis	23 (7.2%)	27 (15.1%)
Jaundice	0 (0%)	95 (53.1%)
Feeding problems	0 (0%)	35 (19.6%)
Respiratory infection	0 (0%)	17 (9.5%)
Others	15 (4.7%)	8 (4.5%)
Total	321 (100%)	179 (100%)

difficulties, and sepsis. kelimesinden sonra. In cases with multiple diagnoses, the primary indication for NICU admission was selected as the principal diagnosis.

Table 5. Primary diagnoses in late NICU admissions according to birth weight groups

Diagnosis	<2500 g (N=35)	2500–3999 g (N=1164)	≥4000 g (N=57)	Total (N=179)
Jaundice	6 (42.9%)	80 (46.8%)	6 (54.5%)	92 (51.4%)
Feeding problems	2 (14.3%)	25 (14.6%)	1 (9.1%)	28 (15.6%)
Sepsis	3 (21.4%)	20 (11.7%)	1 (9.1%)	24 (13.4%)
Hypoglycemia	1 (7.1%)	11 (6.4%)	2 (18.2%)	14 (7.8%)
Respiratory infection	0 (0%)	7 (4.1%)	0 (0%)	7 (3.9%)
Respiratory distress / TTN	1 (7.1%)	6 (3.5%)	0 (0%)	7 (3.9%)
Others	1 (7.1%)	10 (5.8%)	1 (9.1%)	12 (6.7%)
Total	14 (100%)	171 (100%)	11 (100%)	179 (100%)

“In normal birth weight infants, jaundice accounted for the majority of late admissions” cümlesinden sonra.

Türkiye Maternal Fetal Tıp ve Perinatoloji Derneği
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SS-036

İntrapartum İlerleme Açısı ve Baş Perine Mesafesi Ölçümünün Doğum Şeklini ve Süresini Öngörmedeki Yerinin Değerlendirilmesi

Merve Balki Yüzüak[1], Arzu Yavuz[1], Gökçe Naz Küçükbaş Özönder[2]
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OBJECTIVE: To predict the mode of delivery and the duration from the active phase to birth by using the angle of progression and head-perineum distance measurements during intrapartum monitoring.

MATERIAL-METHODS: The labor progression of 128 term pregnant women who entered spontaneous labor was monitored. Cervical dilatation, as determined by vaginal examination at 6 cm and 10 cm, was recorded, and at these time points head–perineum distance (HPD) and angle of progression (AoP) were measured. The measurements together with their corresponding time points were documented, and the role of intrapartum ultrasound in predicting the mode and duration of vaginal delivery was evaluated.

RESULTS: In women who delivered vaginally, the angle of progression (AoP) measured at 6 cm cervical dilatation was wider, and the head–perineum distance (HPD) was longer. In receiver operating characteristic (ROC) curve analysis, an AoP ≤100° and an HPD ≥40 mm were found to be significant predictors of cesarean delivery and operative vaginal delivery. At 10 cm dilatation, AoP was inversely correlated with the duration of the active and second stages of labor, whereas HPD measured at both 6 cm and 10 cm dilatation showed a positive correlation with the duration of the active and second stages.

CONCLUSION: We concluded that the angle of progression (AoP) and head–perineum distance (HPD), as assessed by intrapartum ultrasound, are significant predictors of both the mode and duration of delivery.

Anahtar Kelimeler: Doğum, Doğum eylemi, İntrapartum ultrasonografi, Sezaryen,Vajinal doğum

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SS-037

Postpartum Retroperitoneal Hematoma Managed by Drainage and Arterial Embolization: Two Case Reports

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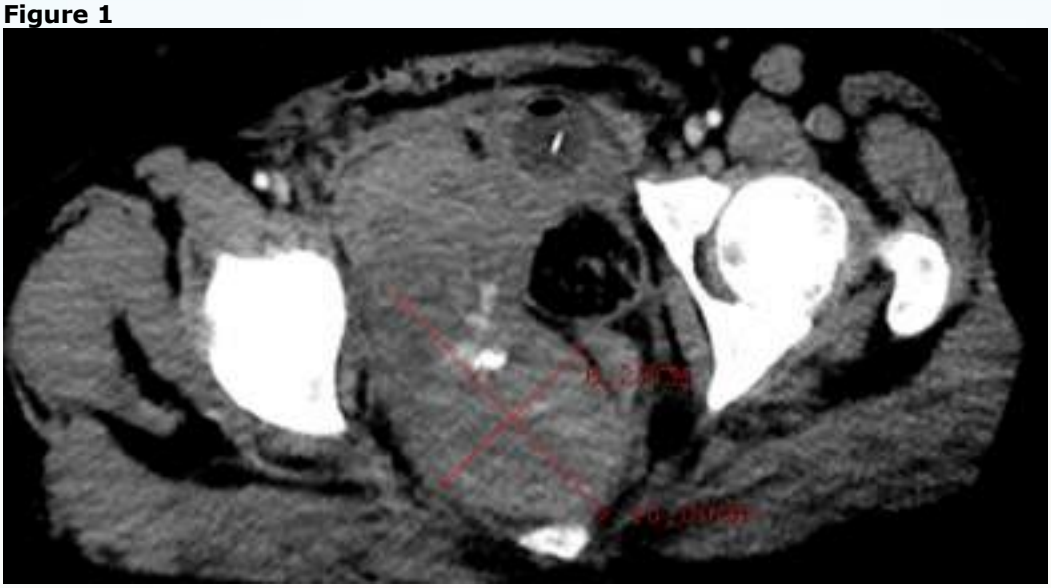
Postpartum hematoma is a rare but potentially life-threatening complication. Clinical presentation varies according to the location of the hematoma. Surgical intervention is generally the initial treatment, whereas laparotomy is indicated when hematoma progression or clinical deterioration occurs. In recent years, arterial embolization has been increasingly recognized as a safe and effective alternative for controlling retroperitoneal bleeding in hemodynamically stable patients. Here, we report two cases of postpartum retroperitoneal hematoma successfully managed in our clinic with a combined approach of surgical drainage and arterial embolization.

The first case involved a patient who developed hemorrhagic shock due to a supralevator hematoma following vaginal delivery. Initial vaginal drainage was performed, but laparotomy was required as the hematoma extended into the supralevator region. Hemodynamic stability was achieved, and selective embolization of the anterior branch of the right internal iliac artery provided definitive hemostasis. The second case occurred after cesarean delivery, in which the patient presented with retroperitoneal hematoma and hemodynamic instability. Laparotomy revealed a hematoma extending from the right lateral Kerr line to the diaphragm, which was drained. Embolization with a particulate agent successfully controlled active bleeding identified during angiography.

These cases highlight the effectiveness of arterial embolization as an adjunct to surgical drainage in the management of postpartum retroperitoneal hematomas. Our experience supports its role as a minimally invasive and effective alternative to traditional surgical techniques in selected patients.

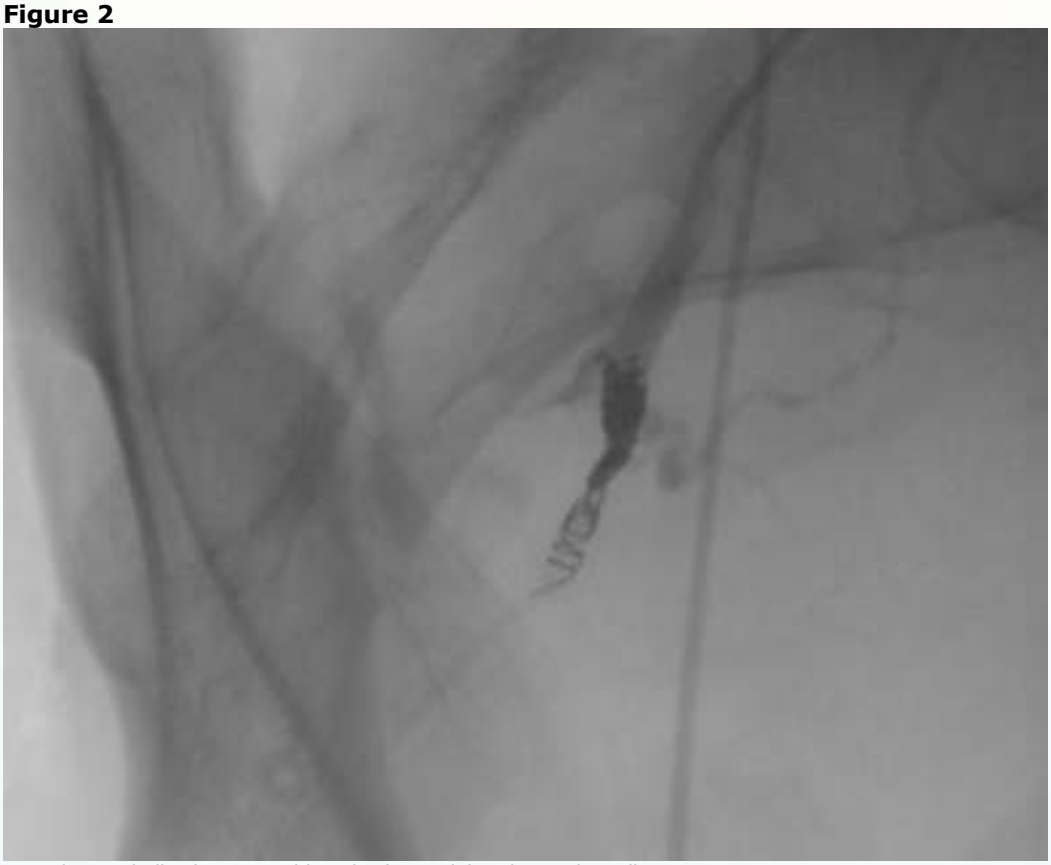
Anahtar Kelimeler: postpartum hematoma, retroperitoneal hemorrhage, arterial embolization

Figure 1



Preoperative computed tomography image of hematoma extending into the retroperitoneum

Figure 2



Complete embolization was achieved using peripheral vascular coils.

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SS-038

Approach to a term patient with loss of consciousness-management of hemorrhagic strokes and eclampsia

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Hemorrhagic strokes in pregnant women have a high morbidity and mortality rate.Strokes are seen in 30/100000 patients with a 6-fold higher risk for patients with hypertensive disorders of pregnancy.Intracranial hemorrhages require an emergent and multidisciplinary intervention.The risk is higher in third-trimester and puerperium, therefore clinical suspicion and timely management is at utmost importance for the diagnosis and treatment of intracranial hemorrhages.A 40 year old nulliparous woman 38 weeks 5 days of gestation presented to our emergency clinic with loss of consciousness and high blood pressure.The day before her admission she complained of vomiting and headache and she was scheduled for cesarean delivery the next day. On initial inspection her Glasgow Coma Scale was 5 out of 15 with decerebrate posture, no initial verbal response and spontaneous eye opening to pain stimuli. Blood pressure was measured 183/109.After initial resuscitation patient was admitted for emergent cesarean delivery.A bolus magnesium infusion was administered preoperatively.4220 gr APGAR 1' 8, 5' 9 girl baby was delivered successfully.Postoperatively a cranial CT showed hemorrhagic densities in 3rd, 4th and lateral ventricles and a 20x10mm intraparenchymal hemorrhage on the right side of nucleus caudatus.Extensive hemorrhage and hydrocephalic dilatation were observed. Cerebral parenchymal and posterior fossa edema was present, cortex and medula distinction was lost.Upon findings patient was transferred to ICU and external ventricular drainage (EVD)was performed by neurosurgery.Patient received 24 hours of magnesium infusion for the prevention of further eclampsia episodes and was put on intravenous nicardipine for regulation of hypertension.Patient gained consciousness after EVD without sequela.

Keywords: Glascow coma scale, emergent approach,hemorrhagic stroke,hypertensive disorder, preeclampsia,

SS-039

A Case of Congenital Syphilis Presenting with Fetal Hepatosplenomegaly

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INTRODUCTION: Treponema pallidum is a sexually transmitted gram-negative spirochete. Perinatal transmission most commonly occurs when the mother is in the primary, secondary, or early latent stage of infection.

CASE: A 31-year-old multigravida woman at 28 weeks of gestation was referred due to suspected fetal microcephaly. Maternal examination and history were unremarkable. Fetal ultrasound revealed hepatosplenomegaly, placentomegaly, oligohydramnios, and growth restriction. Maternal serology showed VDRL-RPR:1/64 and positive TPHA, consistent with late latent syphilis. She received intramuscular benzathine penicillinG, 2.4millionIU weekly for three doses. Amniocentesis (CMV and Toxoplasma PCR-DNA) and cordocentesis (karyotype, microarray, exome panel) were normal. Eight weeks after treatment, placentomegaly, hepatosplenomegaly, oligohydramnios, and growth restriction regressed, and VDRL-RPR titer decreased to 1/8.

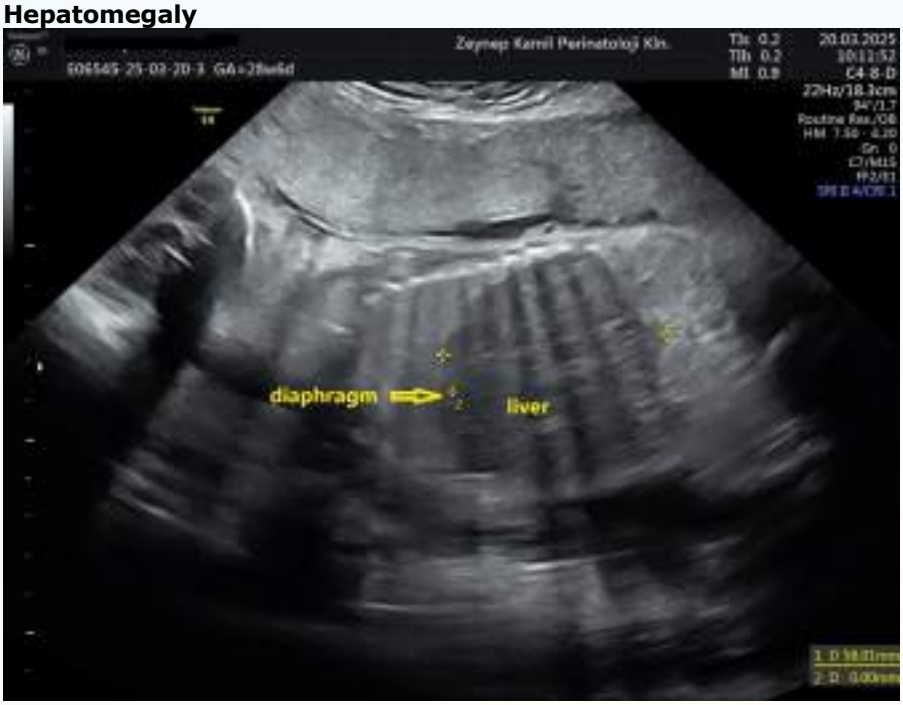
At 38+6weeks, cesarean section was performed for insulin-controlled gestational diabetes and previous C-section. A 3450g female infant with Apgar scores of 8-9 was delivered. Initially asymptomatic, the newborn developed diffuse rash (including palms and soles), thrombocytopenia, and suspected periostitis within one week. Neonatal serology showed VDRL-RPR:1/4 and positive TPHA. According to CDC criteria, the infant was diagnosed with "highly probable congenital syphilis" and treated with intravenous crystalline penicillin G (50,000U/kg).

DISCUSSION: Fetal signs usually appear after 16–20 weeks when immune competence develops. Hepatomegaly is the earliest and most common manifestation. Without treatment, risks of abortion, morbidity, and mortality are high. PenicillinG is the only proven therapy, though congenital syphilis may still occur in some cases.

CONCLUSION: Syphilis should be considered in the differential diagnosis of fetal hepatosplenomegaly. Early screening and treatment can prevent vertical transmission and complications.

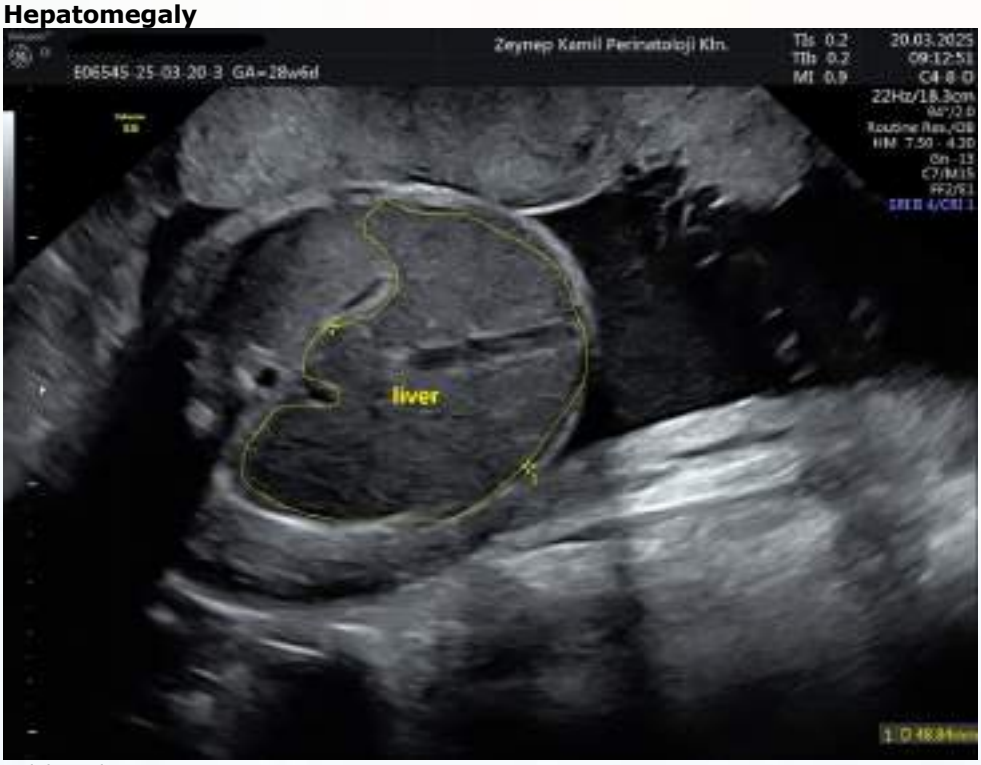
Keywords: congenitalsyphilis, fetalhepatosplenomegaly, benzathinepenicillinG, placentomegaly

Hepatomegaly



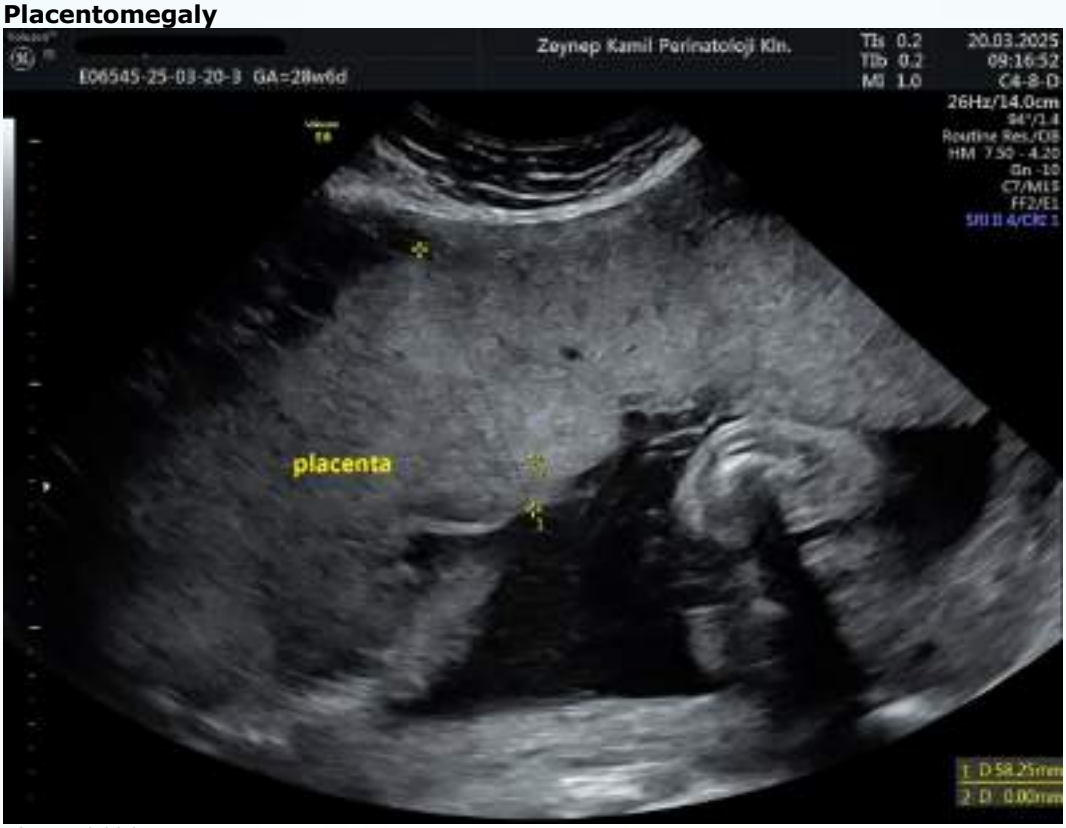
Longitudinal right parasagittal plane right lobe SI length: 58 mm, >95.percentile (Tongprasert F, et al. Normal length of the fetal liver from 14 to 40 weeks of gestational age. J Clin Ultrasound 2011, 39(2): 74–7.)

Hepatomegaly



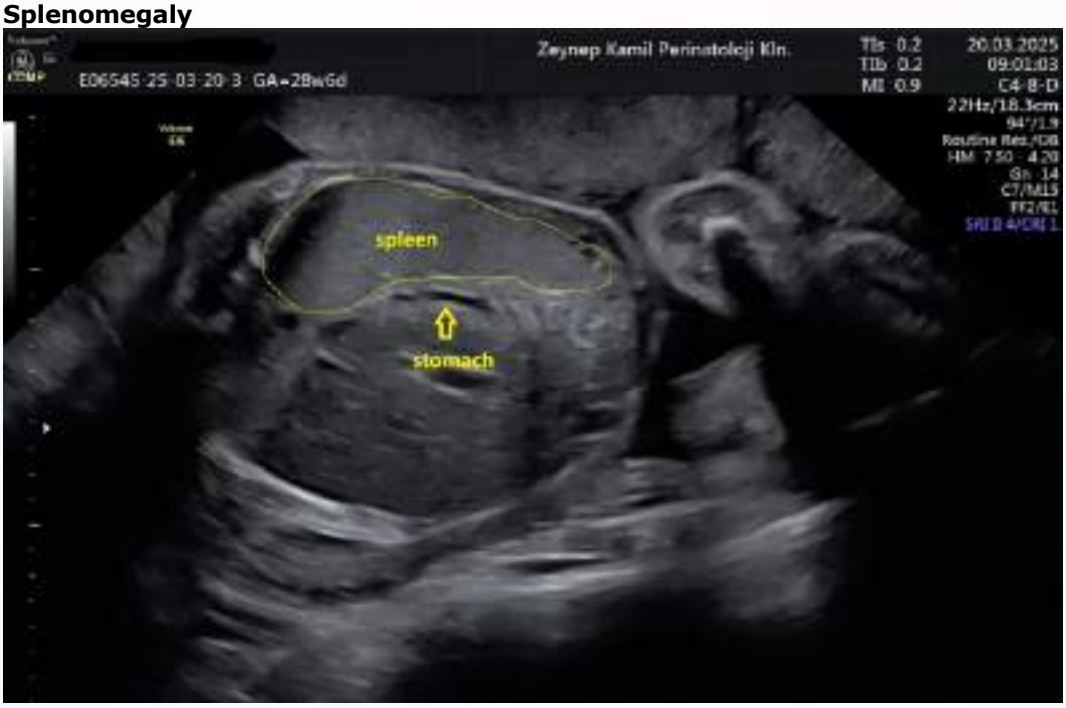
Axial Section

Placentomegaly



Placental thickness: 58 mm

Splenomegaly



Splenic circumference values: 145 mm, >95.percantile (Srisupundit K, et al. Reference range of fetal splenic circumference from 14 to 40 weeks of gestation. Arch Gynecol Obstet 2011; 283(3): 449–53)

SS-040

Sonographic depiction of fetal cardiac failure associated with intrauterine growth restriction due to maternal propylthiouracil use

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BACKGROUND: Hyperthyroidism complicates approximately 0.05-0.2% of pregnancies. Propylthiouracil (PTU), easily crossing the placenta, may be an option for the treatment of symptomatic patients. However, prolonged or high-dose use may lead to fetal hypothyroidism and goiter, which may indirectly increase the risk of fetal hydrops and intrauterine growth restriction (IUGR).

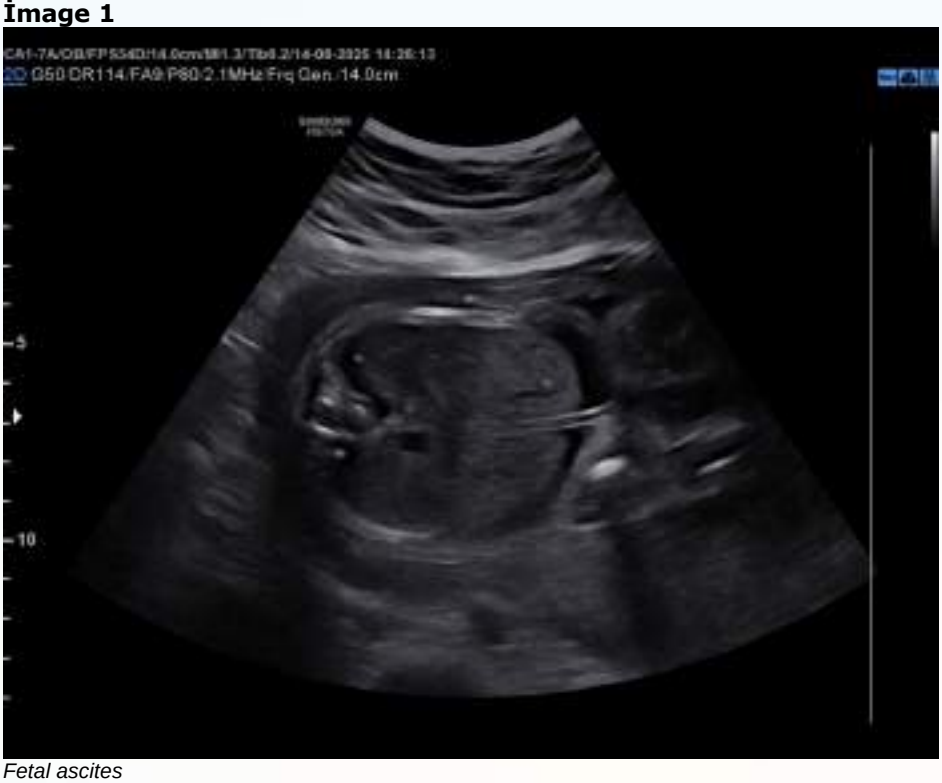
Case Presentation: A 29-year-old primigravida at 29th gestational week was suspected of having fetal IUGR at the routine prenatal visit. She had a history of asymptomatic low thyroid stimulating hormone (TSH), treated with PTU 50 mg/day for 1.5 months prior to admission. Ultrasound examination revealed an IUGR fetus with ascites and cardiomegaly. Superior and inferior vena cava were dilated and tricuspid regurgitation was observed. Doppler studies of the fetus revealed reversed end-diastolic flow in umbilical artery, reversed "a" waves in ductus venosus and pulsations in umbilical vein. These findings suggested decompensated fetal cardiac failure associated with IUGR, probably secondary to maternal PTU use.

Management and Outcome: Antenatal corticosteroids and neuroprotective magnesium sulfate were administered. Emergency cesarean delivery resulted in a 1000 g male neonate with severe bradycardia requiring immediate intubation and transfer to the neonatal intensive care unit. Cord blood gas analysis showed mild acidosis (pH 7.122, BE -12.2). Placental pathology revealed infarctions consistent with intrauterine hypoxia. Newborn thyroid tests demonstrated overt hypothyroidism (TSH: 23.6 mU/L, fT4: 0.42 ng/dL), which was normalized gradually with replacement therapy.

CONCLUSION: Maternal hyperthyroidism requires careful evaluation before initiating antithyroid therapy. Unnecessary treatment of asymptomatic mothers may cause severe fetal hypothyroidism, IUGR and life-threatening fetal complications.

Keywords: Intrauterine Growth Restriction, Hyperthyroidism, Propylthiouracil

İmage 1

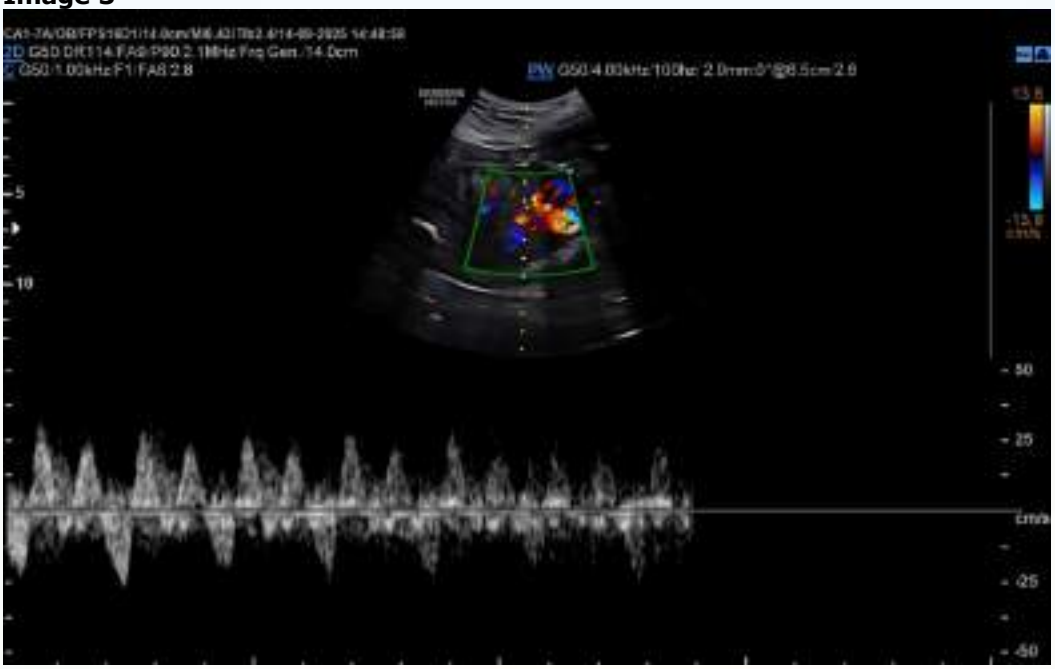


Fetal ascites

İmage 2

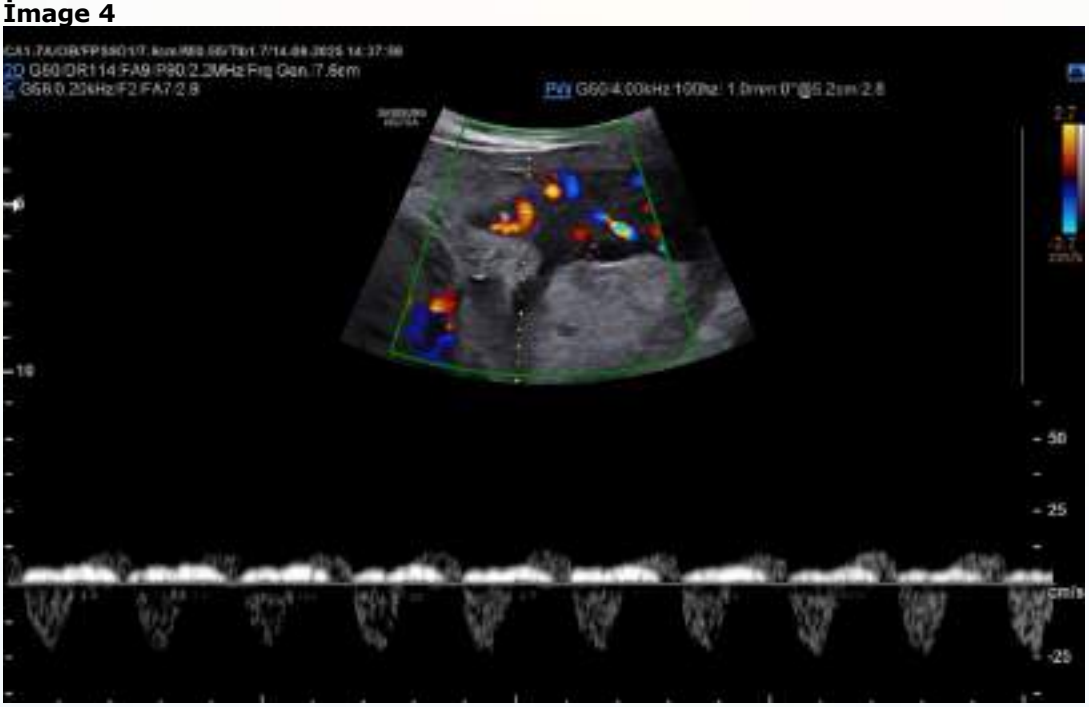
Reversed end-diastolic flow in umbilical artery

İmage 3



Reversed “a” waves in ductus venosus

İmage 4



Pulsations in umbilical vein

SS-041

Evaluation of Serum Inflammatory Markers in Pregnant Women Diagnosed with Class A1 Gestational Diabetes Mellitus

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Department of Perinatology,Uşak Training and Research Hospital, Uşak, Turkey

INTRODUCTION: Gestational diabetes mellitus (GDM) is a condition that can affect fetal and maternal health. Identifying prognostic indicators for GDM may improve risk assessment and patient selection for intensive monitoring. Considering the inherent sensitivity of inflammatory parameters to other factors, studies have shown that neutrophil-lymphocyte ratio (NLR), platelet-neutrophil ratio, platelet-lymphocyte ratio (PLR) can be used in combination as theoretically more reliable markers than individual cells to assess inflammatory status, and increased PLR and NLR have been shown to be independent predictors of GDM. The aim of this study was to evaluate whether systemic inflammatory indices have an impact on adverse pregnancy outcomes in GDM A1 and normoglycemic patients.

Material- METHOD: This retrospective study was conducted at Uşak Training and Research Hospital between 2022 and 2024, and included 40 untreated pregnant women diagnosed with gestational diabetes mellitus and 40 healthy pregnant controls. Pregnancies with multiple gestations, anemia, hematological disorders, inflammatory conditions, history of malignancy, infectious diseases, as well as fetal chromosomal or structural anomalies were excluded from the study.

RESULTS: At the time of birth, white blood cell count, neutrophil count, and neutrophil-to-lymphocyte ratio (NLR) were higher in the GDM group, but no statistically significant difference was found. The complication rate was higher in the GDM group, and the postnatal neonatal observation period was significantly longer.In our study, the failure to predict adverse outcomes despite the high levels of systemic inflammatory markers in the GDM group was thought to be related to the small number of participants.

Keywords: gestational diabetes mellitus, inflammatory marker, complication

Baseline characteristics of participants

	GDM group (n=40)	Non- GDM group (n=40)	P value
Maternal age(years)	28(7)	25 (8)	0.015
Nulliparity(n, %)	6	20	0.001
Female fetal gender (n,%)	19	18	0.823
Gestational age at birth (week)	38 (2)	38 (4)	0.950
Birth weight (gram)	3312 (663)	3010 (980)	0.325
Emergency cesarean section (n,%)	8	2	0.040
APGAR score at 1st minute	9 (1)	9 (1)	0.794
APGAR score at 5th minute	10 (1)	9 (1)	0.819
NICU admission (n, %)	10	6	0.264
Complication (n)	12	4	0.025
24 hour observation (n)	21	6	<0.001

Blood cell counts and inflammatory parameters

	GDM group (n=40)	Non-GDM group (n=40)	P value
WBC (10^9 /L)	12.5 (4.5)	10.6 (3.3)	0.357
Neutrophil (10^9 /L)	9.7 (4.2)	8.4 (2.9)	0.256
Lymphocyte (10^9 /L)	1.590 (0.7)	1.72(0.5)	0.820
Monocyte (10^9 /L)	0.6 (0.2)	0.5 (0.2)	0.600
SII	1042 (659)	1045.3 (531.9)	0.638
SIRI	3.3 (2.2)	2.81 (2.1)	0.474
AISI	613.5 (468)	594 (518)	0.869
NLR	5.6 (3)	5.0 (2.7)	0.109
HG	11.0 (1.7)	10.7 (2)	0.682
PLT	187 (53)	210 (67)	0.064

Note: P-values < 0.05 were considered statistically significant. AISI, aggregated index of systemic inflammation; NLR, neutrophil-to-lymphocyte ratio; SII, systemic inflammation index; SIRI, systemicinflammatory response index

SS-042

Post-Partum Retroperitoneal Hematoma: A Case Managed With Conservative Approach

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OBJECTIVE: Retroperitoneal hematoma primarily occurs as a result of birth trauma, vaginal lacerations, vascular injuries, coagulopathy, and surgical interventions. In this presentation, we will share our experiences regarding the clinical course of retroperitoneal hematoma cases occurring in the postpartum period, findings detected by imaging methods, and the follow-up process accompanied by conservative treatment.

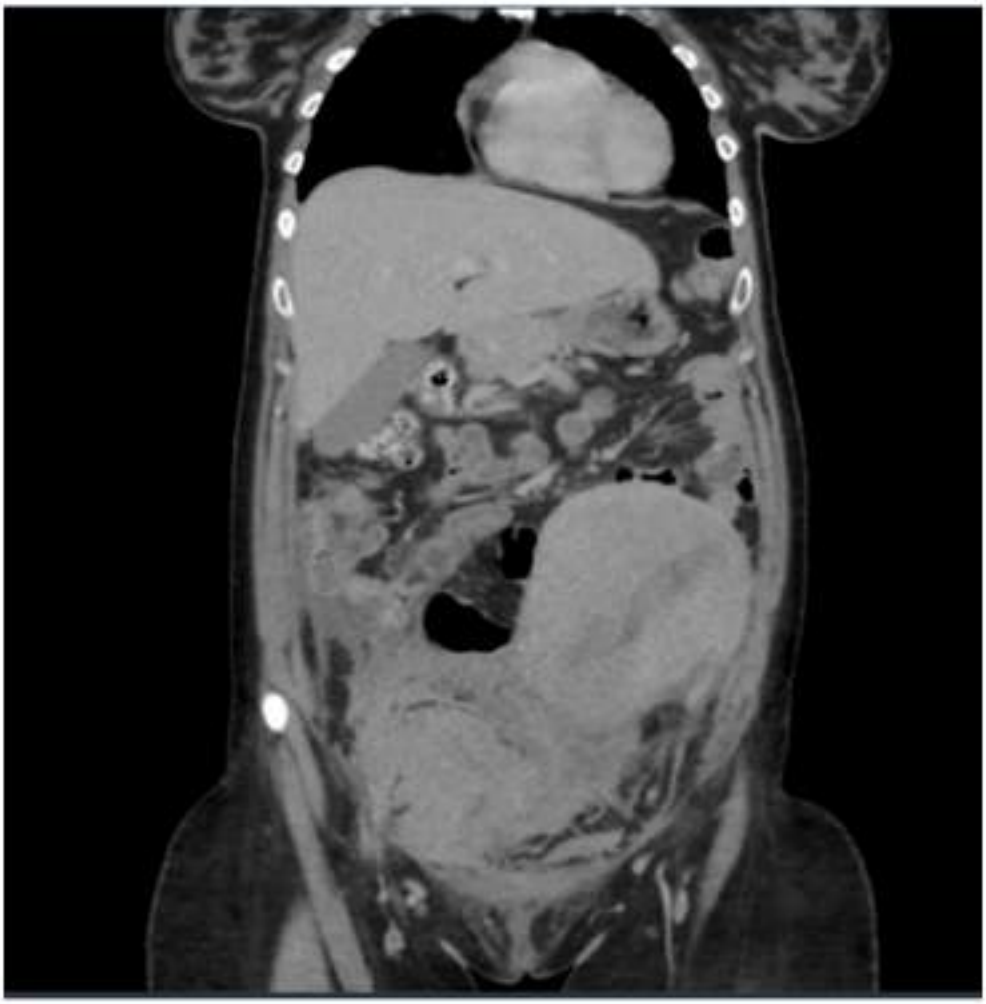
METHOD: We aimed to present the case using tomographic sections.

FINDINGS: A 28-year-old primigravida who delivered vaginally at an external center was referred to our clinic with abdominal pain. Laboratory evaluation revealed severe anemia (Hb 6.8 g/dL), leukocytosis (17.61×10³/μL), elevated C-reactive protein (27 mg/L), and normal coagulation parameters. Abdominal computed tomography demonstrated a retroperitoneal organized hematoma measuring 180×94 mm in the right iliac fossa, adjacent to iliac vessels, compressing the uterus and bladder (Figure 1). The patient received red blood cell and fresh frozen plasma transfusions, initially combined with ceftriaxone and clindamycin, later adjusted to piperacillin/tazobactam. Given hemodynamic stability, conservative management was preferred over surgical intervention. During 21 days of hospitalization, follow-up ultrasonography demonstrated hematoma regression (126×92 mm). The patient was discharged in stable condition. At 11-month follow-up, ultrasonography confirmed complete resolution of the para-iliac hematoma, supporting successful non-operative management of this rare postpartum complication.

RESULT: Retroperitoneal hematoma is a rare condition in the postpartum period, but it can lead to clinically significant complications. Conservative treatment and close clinical follow-up can yield successful outcomes without the need for surgical intervention.

Keywords: Spontaneous vaginal delivery, Postpartum retroperitoneal hematoma, Conservative treatment

Retroperitoneal hematoma abdominal CT scan image



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SS-043

Prenatal diagnosis of Turner syndrome with multiple fetal anomalies

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INTRODUCTION: Turner syndrome occurs in approximately one in 2,500 live female births, representing one of the most common chromosomal abnormalities compatible with survival. Cystic hygroma appears in 75-80% of cases during first and second trimesters, serving as a key sonographic marker. The combination of multiple structural anomalies including cardiac defects, pleural effusions, and limb deformities significantly increases the likelihood of chromosomal abnormalities, particularly monosomy X.

CASE: A 24-year-old woman, gravida 2, para 1, presented for routine prenatal care at fifteen weeks gestation. Ultrasonographic examination revealed multiple fetal anomalies including cystic hygroma (Figure 1), pleural effusion, pericardial effusion, atrioventricular septal defect, intracardiac hyperechogenic focus (Figure 2), hyperechogenic bowel, right foot pes equinovarus, and single umbilical artery.

Following comprehensive genetic counseling regarding chromosomal abnormality risk, the family accepted prenatal diagnostic testing. Chorionic villus sampling confirmed 45,X0 karyotype consistent with Turner syndrome. The patient underwent medically induced termination without complications and was discharged on postoperative day two. Postnatal skin biopsy chromosomal analysis confirmed the Turner syndrome diagnosis.

CONCLUSION: This case demonstrates the importance of comprehensive ultrasonographic evaluation when multiple fetal anomalies are detected. Early recognition of characteristic sonographic patterns associated with Turner syndrome enables timely genetic counseling and informed decision-making regarding pregnancy management. The combination of multiple structural anomalies including cardiac defects, pleural effusions, and limb deformities significantly increases the likelihood of chromosomal abnormalities, particularly monosomy X.

Keywords: Cystic hygroma, fetal anomalies, prenatal diagnosis, Turner syndrome, ultrasonography.

Figure 1.Prenatal ultrasonographic findings of cystic hygroma (A)Axial view demonstrating bilateral cystic hygroma (arrow)in the nuchal region. (B)Coronal view showing the characteristic septated cystic structure(arrow) consistent with cystic hygroma

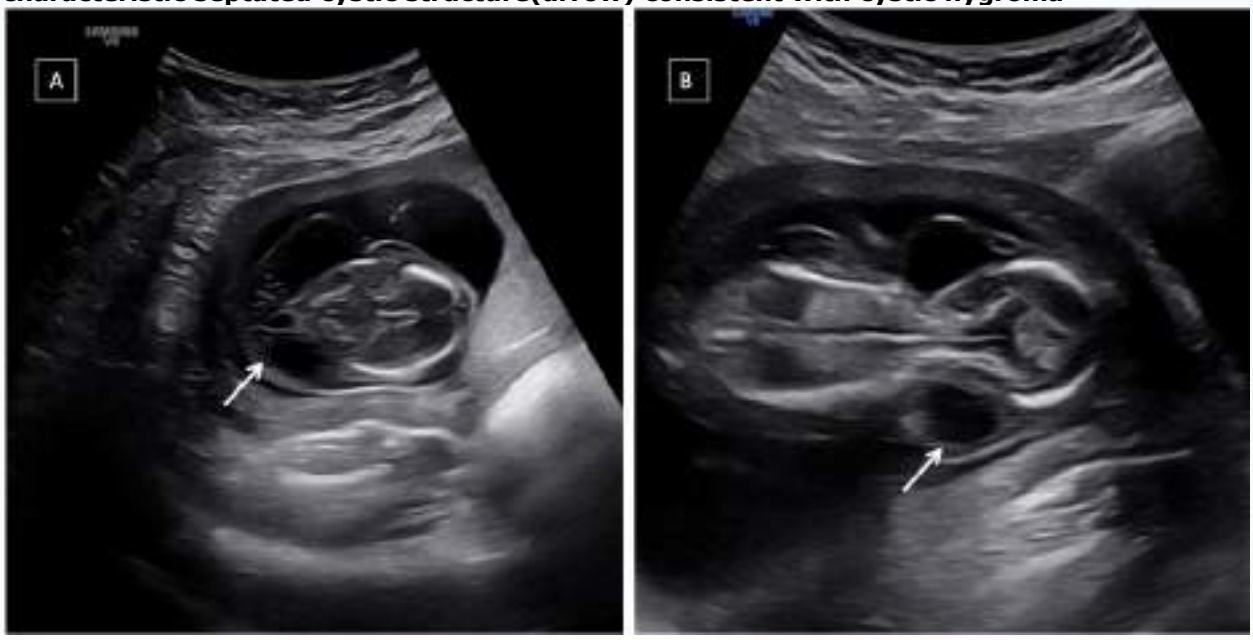


Figure 2. (A) Four-chamber view demonstrating atrioventricular septal defect(star) and bilateral pleural effusion (arrows). (B) Sagittal view demonstrating pleural effusion (arrows), pericardial effusion (cross) and intracardiac hyperechogenic focus.

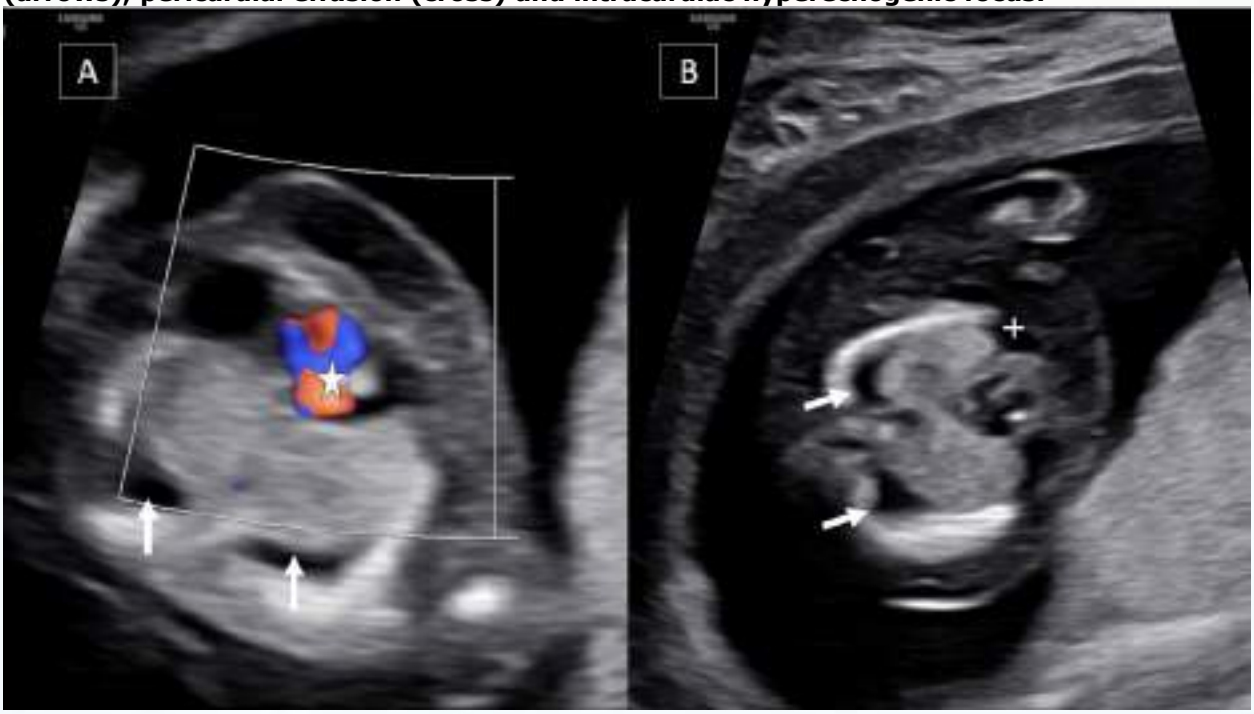
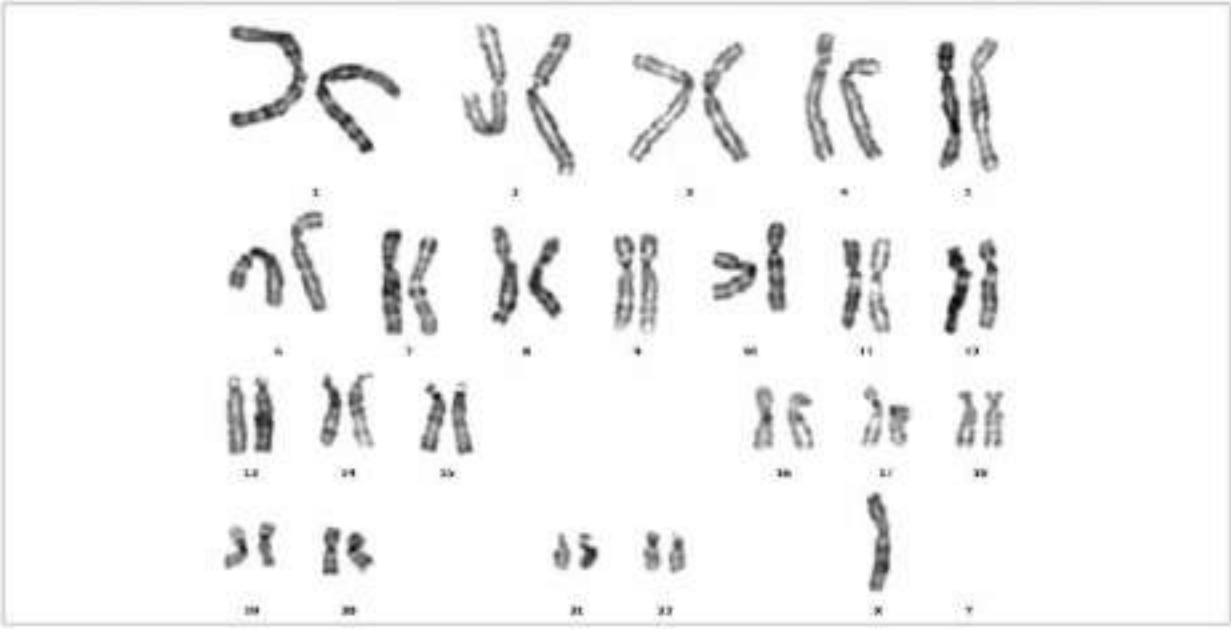


Figure 3. Karyotype analysis showing 45,X0 Turner syndrome confirmed by chorionic villus sampling.



SS-044

A Successful Pregnancy with Wolfram Syndrome: A Case Report

Esra Nuhoğlu Bilgin[1], Pınar Tokdemir Çalış[1], Hatice Aydın Salman[1], Arzu Sümer Ortaç[1], Esin Şahin Toruk[1], Bilal Emre Erzeneoğlu[1], Tural Mammadaliyev[2], Deniz Karçaaltıncaba[1]
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[2]Department of Neonatology, Medical Faculty of Gazi University, Ankara, Turkiye

Wolfram syndrome (DIDMOAD) is a rare autosomal recessive neurodegenerative disorder characterized by juvenile-onset diabetes mellitus, optic atrophy, diabetes insipidus, and sensorineural hearing loss. Fertility is often impaired, but although it ıs rare, successful pregnancies have been described.

CASE: We present the case of a 38-year-old woman with Wolfram syndrome carrying a WFS1 mutation. She had a history of type 1 diabetes mellitus since the age of 5, diabetes insipidus since age 13, and optic atrophy. Despite two prior pregnancy losses, she achieved a successful pregnancy with close multidisciplinary follow-up. At 36 weeks, she delivered a healthy male infant via cesarean section.

CONCLUSION: Although rare, pregnancy in Wolfram syndrome is feasible. Multidisciplinary management, strict glycemic control, and genetic counseling are essential to optimize outcomes.

Keywords: Wolfram syndrome, DIDMOAD, pregnancy, diabetes mellitus, optic atrophy, case report

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SS-046

Emanuel syndrome in a patient with Neurofibromatosis Type 1: Management of two consecutive pregnancies

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[1]Sakarya University Faculty of Medicine, Department of Obstetrics and Gynecology, Division of Perinatology

[2]Sakarya Training and Research Hospital, Medical Genetics Unit

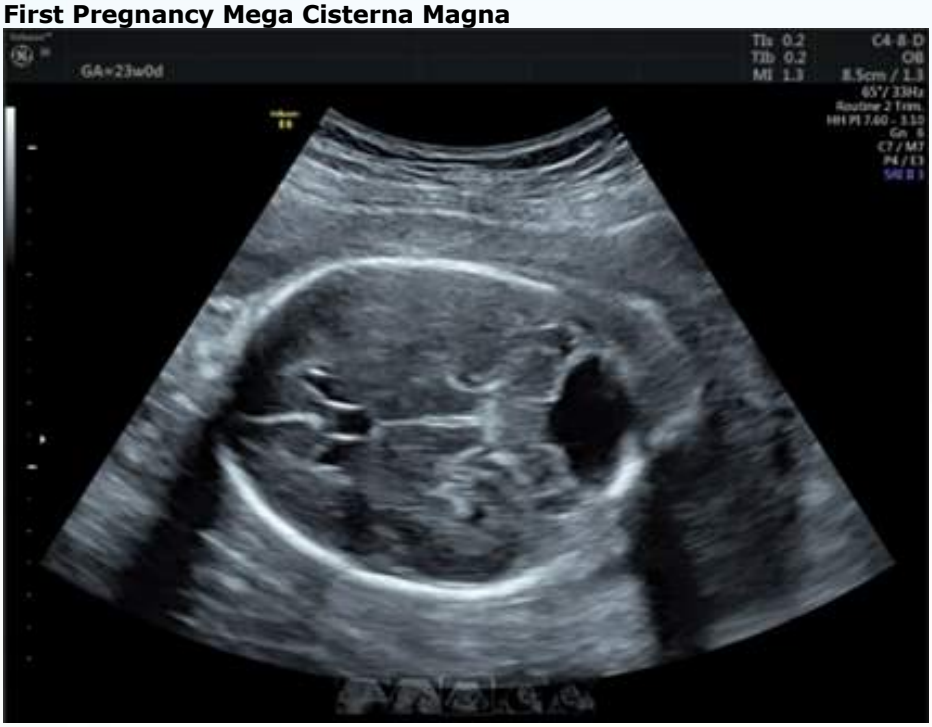
OBJECTIVE: This case report details the prenatal diagnosis and management of two consecutive pregnancies complicated by Emanuel syndrome in a 23-year-old patient with Neurofibromatosis Type 1 (NF1). The objective is to highlight the critical role of genetic counseling, advanced prenatal imaging, and cytogenetic testing in managing families with balanced chromosomal translocations, emphasizing a multidisciplinary approach to prenatal care.

CASE: The patient, a known NF1 carrier, was diagnosed as a balanced carrier of the t(11;22)(q23;q11) translocation following genetic evaluation after her second pregnancy. Ultrasound examination at 21 weeks gestation revealed anhydramnios, nuchal fold thickening (7.3 mm), mega cisterna magna, a ventricular septal defect, and bilateral multicystic dysplastic kidneys. Amniocentesis with chromosomal microarray analysis confirmed Emanuel syndrome, resulting in medical termination. In her subsequent pregnancy, second-trimester ultrasound identified Dandy-Walker malformation and arachnodactyly. Repeat amniocentesis confirmed recurrent Emanuel syndrome, leading to another termination. Postmortem examination revealed additional features including sacral dimple, multiple neurofibromas, and arachnodactyly.

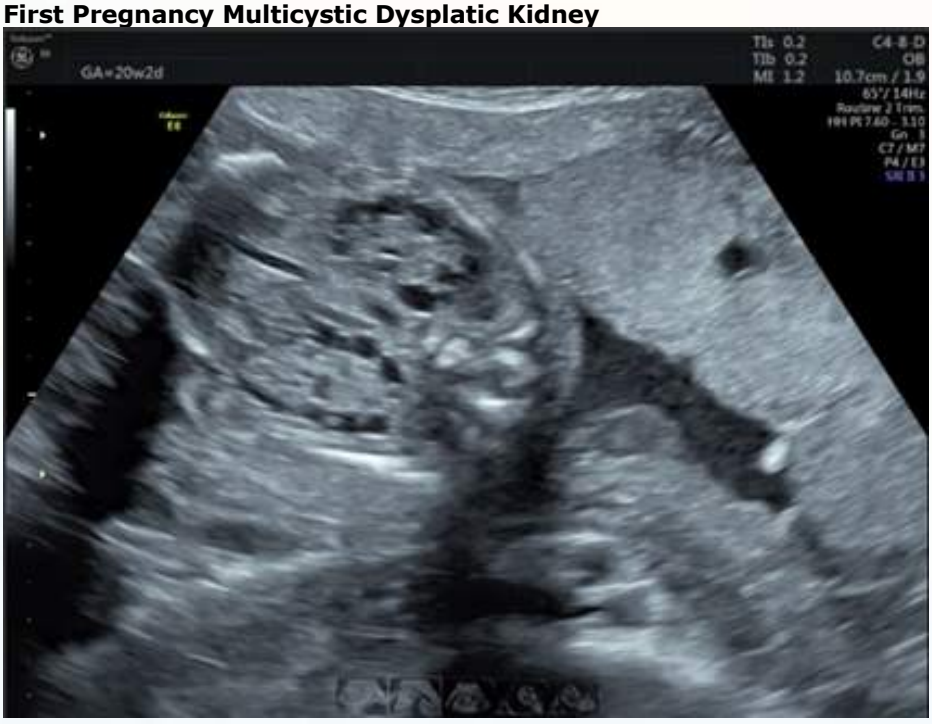
CONCLUSION: This case underscores the significant recurrence risk associated with balanced translocations and demonstrates the necessity of comprehensive genetic counseling. Emanuel syndrome presents with severe neurodevelopmental impairment and multiple congenital anomalies. Preimplantation genetic diagnosis represents a crucial option for future pregnancies. A coordinated multidisciplinary approach involving perinatology, genetics, and pediatrics is essential for optimal management. Early prenatal screening and advanced genetic testing remain fundamental components for informed decision-making and improved reproductive outcomes in high-risk families.

Keywords: Emanuel Syndrome, Neurofibromatosis Type 1, Pregnancy

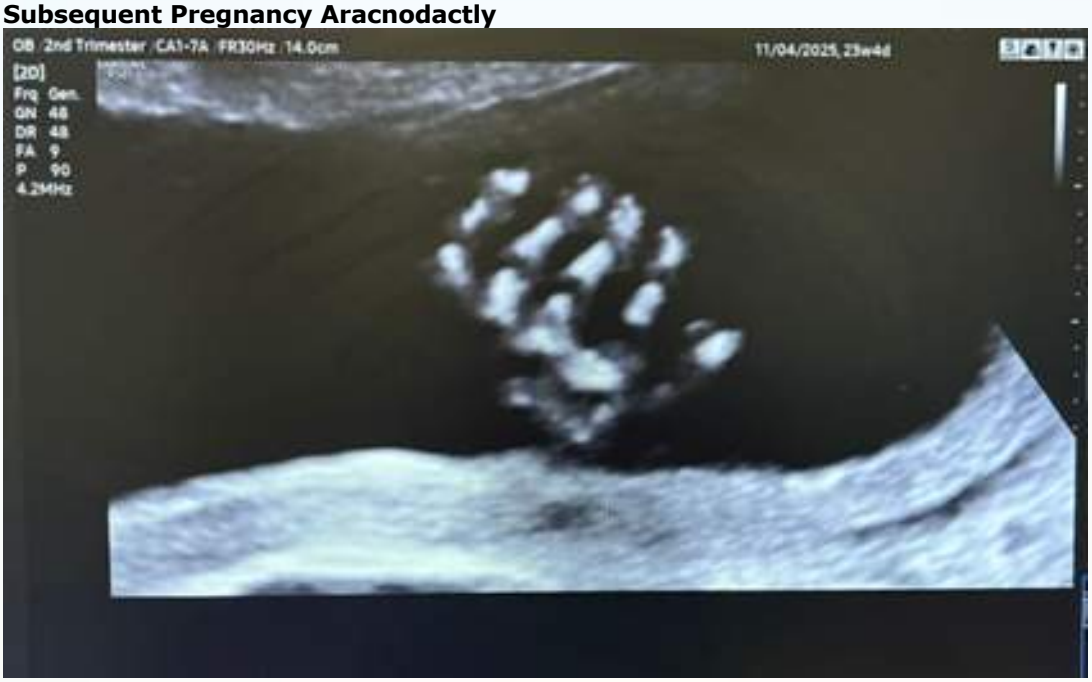
First Pregnancy Mega Cisterna Magna



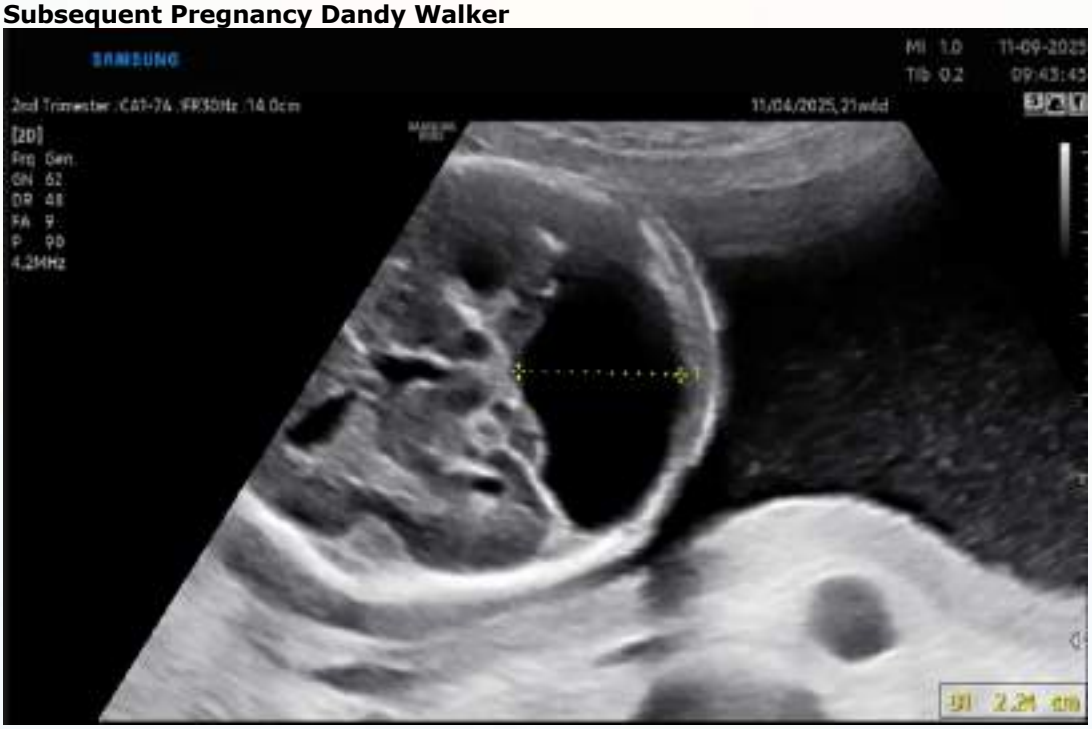
First Pregnancy Multicystic Dysplatic Kidney



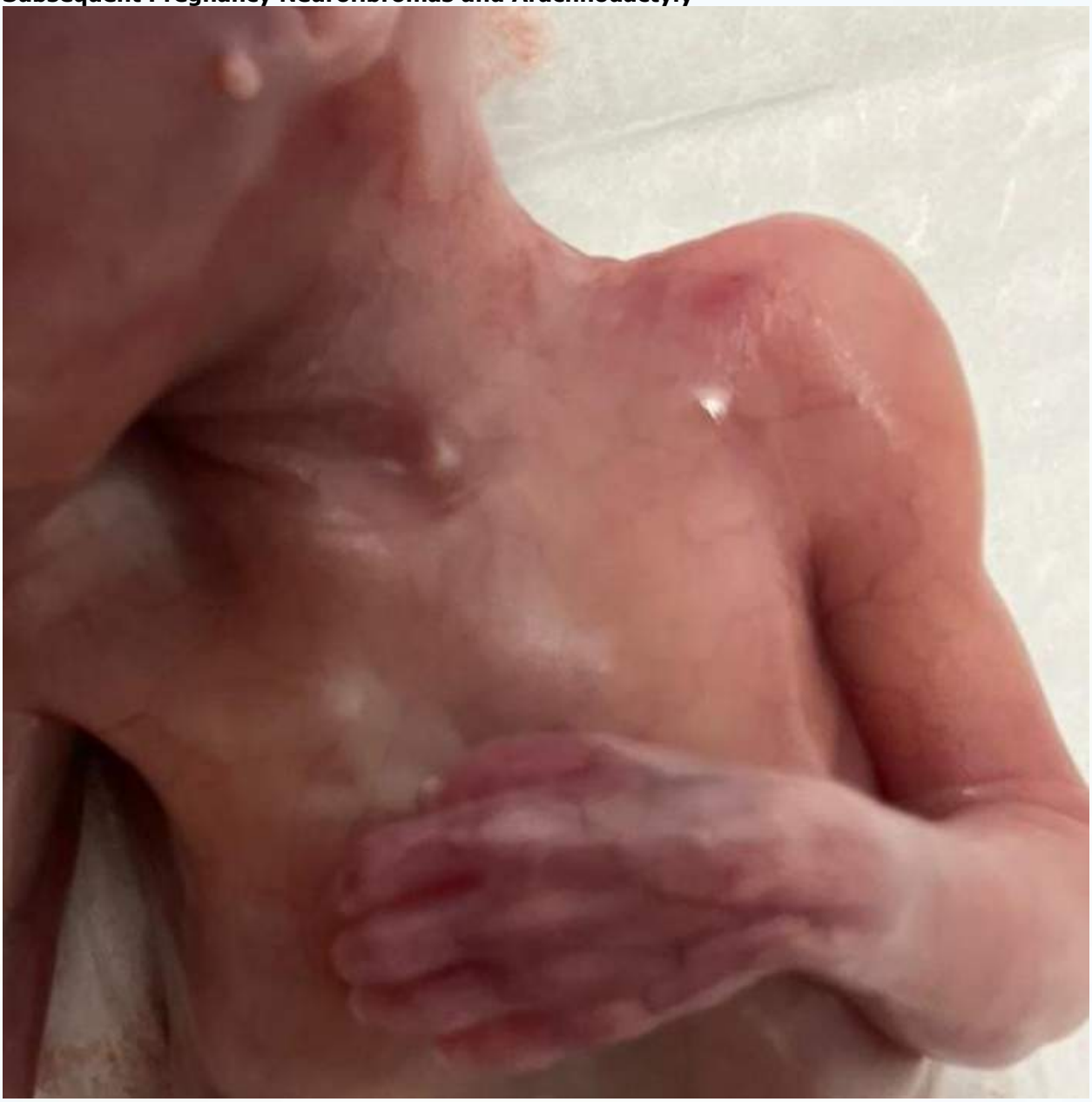
Subsequent Pregnancy Aracnodactly



Subsequent Pregnancy Dandy Walker



Subsequent Pregnancy Neurofibromas and Arachnodactyly



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SS-047

Prenatal Diagnosis and Postnatal Outcome of 3 Cases with Fetal Cholelithiasis: A Case Series

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INTRODUCTION: Fetal cholelithiasis is a rare third-trimester sonographic finding, usually benign and self-limiting. Reported maternal and fetal risk factors include diabetes, hemolytic disorders, congenital anomalies, drug exposure, and twin pregnancies. We present three cases of prenatally diagnosed fetal cholelithiasis with postnatal follow-up.

Cases: A primigravida with hypothyroidism, diet-controlled gestational diabetes, and IVF conception was diagnosed with gallbladder sludge at 37 weeks. Vaginal delivery at 38+3 weeks resulted in a 3130 g male. Ultrasound on postnatal day 8 showed a normally appearing gallbladder without cholelithiasis. A gravida 3 with epilepsy and prior bariatric surgery had a small apical ventricular septal defect and early growth restriction detected during the second-trimester scan. At 32 weeks, ultrasound revealed multiple gallstones. Cesarean delivery at 37 weeks resulted in a 2270 g female. The neonate required NICU admission for TTN and sepsis. Ultrasound on day 1 showed sludge, which had resolved by the two-month follow-up.

A primigravida with FMF carrier was referred in the second trimester for suspected aortic coarctation, later confirmed as mild. At 38 weeks, ultrasound demonstrated gallstones. Vaginal delivery at 40+1 weeks resulted in a 3030 g male. Ultrasound on day 2 showed sludge, while follow-up at 6 months and 1 year confirmed complete resolution without clinical symptoms.

CONCLUSION: All three fetuses were asymptomatic with spontaneous resolution, consistent with literature reporting >90% resolution within the first year. This series highlights the importance of prenatal detection, parental counseling, and postnatal surveillance. Although generally benign, awareness of risk factors and rare persistent cases remains essential.

Keywords: Fetal cholelithiasis,Prenatal diagnosis, Perinatal outcome

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SS-048

A Rare Case of Prenatal Multisystem Anomaly Diagnosed Postnatally with FANCF Mutation and Accompanying PEX7 Variant

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Fanconi Anemia (FA) is a rare autosomal recessive syndrome characterized by chromosomal instability. This presentation describes a case with prenatal multisystem anomalies in which a homozygous FANCF mutation and a heterozygous PEX7 variant were identified postnatally.

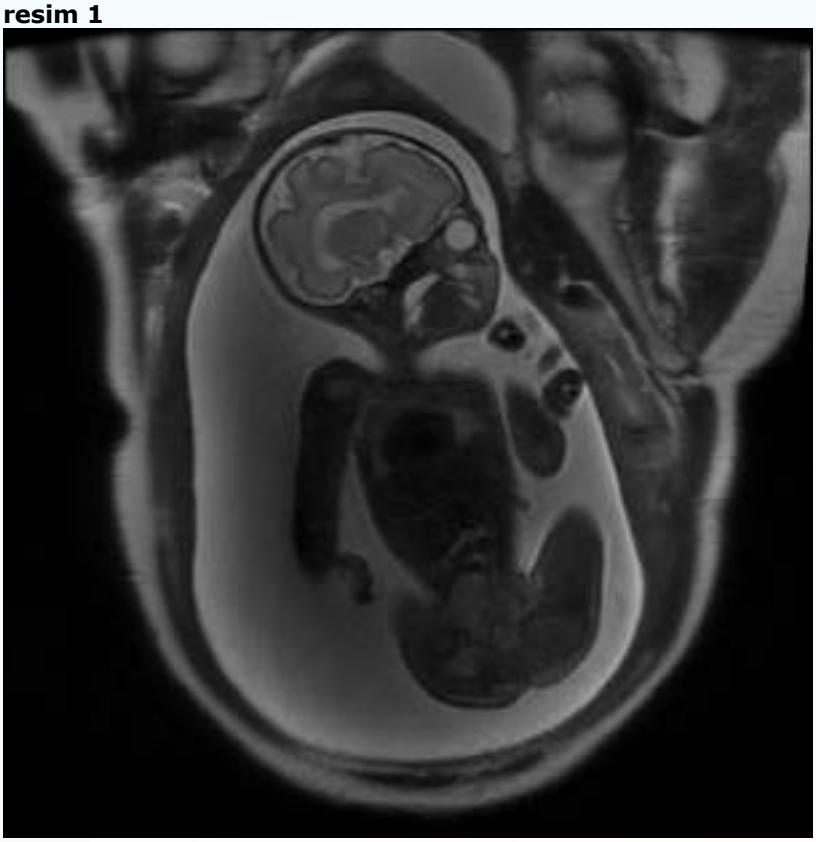
A 39-year-old primigravida conceived. At 30+3 weeks of gestation, prenatal ultrasound revealed duodenal atresia (double bubble sign), bilateral radial agenesis, oligodactyly, clenched hand deformities, and fused pelvic kidneys. Polyhydramnios was present. Fetal echocardiography showed dilation of the right atrium and ventricle, increased right ventricular wall thickness, and minimal pericardial effusion. Postnatal examination confirmed the diagnosis of anal atresia. Whole exome sequencing (WES) identified a homozygous FANCF mutation NM_022725.3:c.484_485del (p.Leu162fs) and a heterozygous PEX7 variant NM_000288.3:c.370_396del (p.Gly124_Ser132del). There was no history of consanguinity in the family.

The coexistence of duodenal and anal atresia, fused kidneys, bilateral upper limb anomalies, and signs of cardiac remodeling suggested a syndromic condition. WES confirmed the diagnosis of Fanconi Anemia (FA). FANCF is a core protein involved in maintaining the stability of the FA pathway complex, and its dysfunction leads to multisystem involvement. Although the accompanying PEX7 variant may not directly contribute to the phenotype, it could have aggravated the clinical presentation as a modifier gene.

This case highlights the importance of advanced imaging techniques for prenatal diagnosis and the role of postnatal genomic analyses in identifying rare genetic syndromes. FANCF mutations may not always present with classical FA features prenatally; therefore, molecular analyses such as WES are essential for definitive diagnosis. Identifying coexisting variants can further elucidate genotype-phenotype correlations.

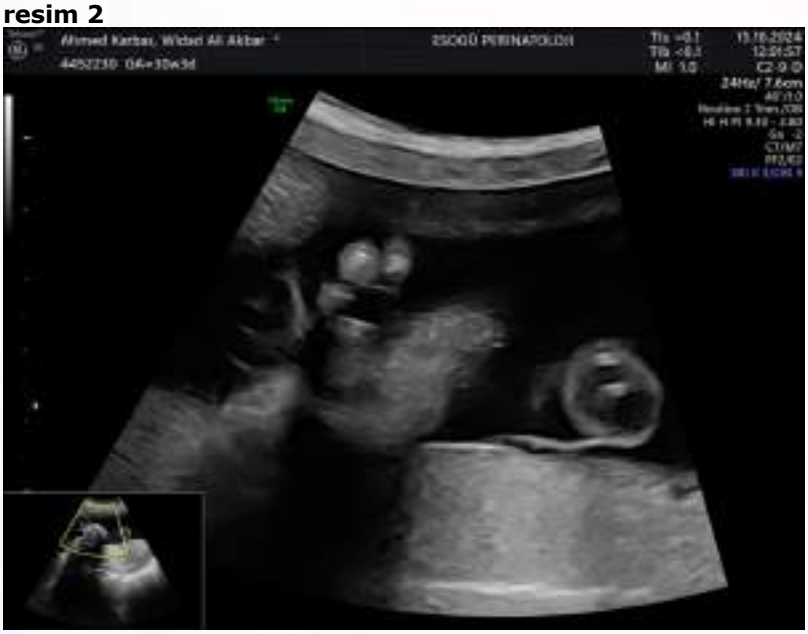
Anahtar Kelimeler: FANCF, PEX7, fused kidney, WES

resim 1



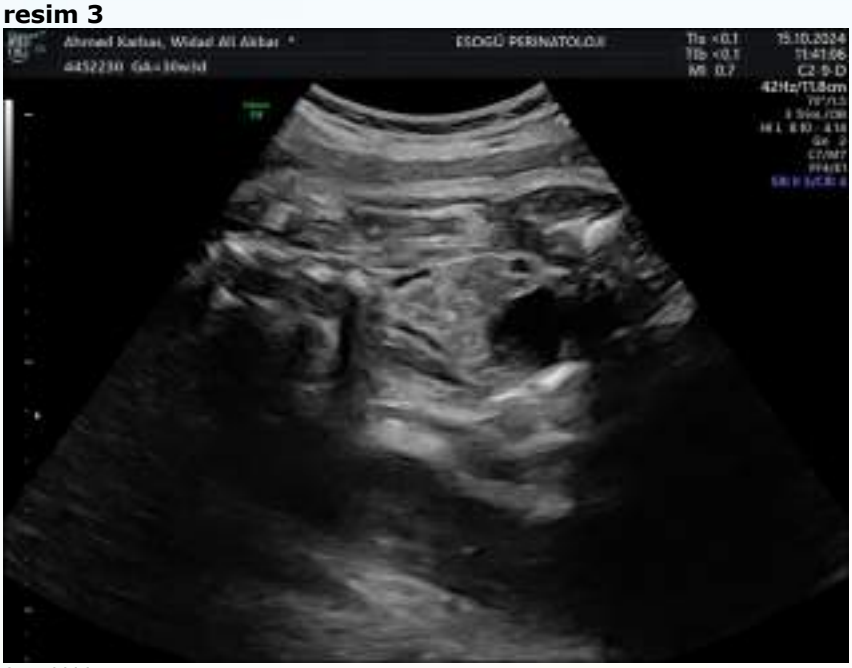
clench hand, radius agenezisi

resim 2



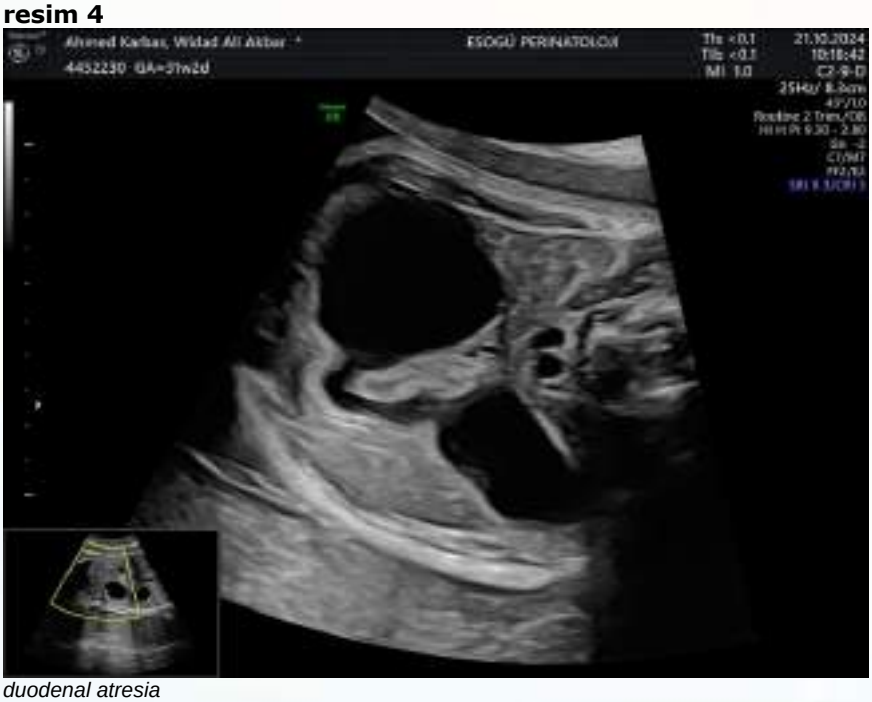
Oligodaktili

resim 3



fused kidneys

resim 4



duodenal atresia

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The Diagnostic Value of Placental Leukemia Inhibitory Factor (LIF) and Vascular Endothelial Growth Factor (VEGF) Levels and Their Combined Use in the Prediction of Preeclampsia

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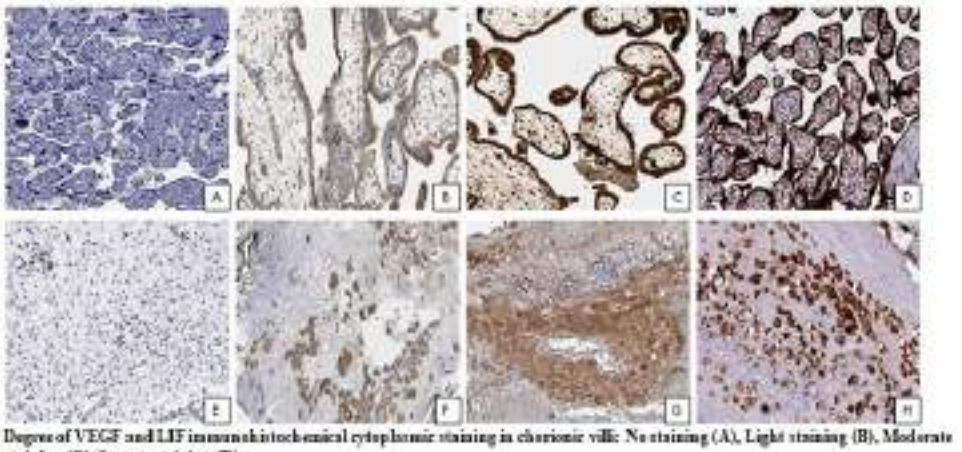
INTRODUCTION: Preeclampsia is a complex multisystemic disorder characterized by new-onset hypertension and organ dysfunction during pregnancy. It remains a leading cause of maternal and fetal morbidity and mortality worldwide. Early detection of preeclampsia is critical to reduce adverse outcomes, yet there is currently no gold standard screening test. Leukemia Inhibitory Factor (LIF) and Vascular Endothelial Growth Factor (VEGF) are crucial regulators of placental development and angiogenesis, processes often disrupted in preeclampsia. This study aims to evaluate the effectiveness of placental LIF and VEGF expression as predictive biomarkers for preeclampsia.

METHODS: A prospective cohort study was conducted including 60 pregnant women: 40 diagnosed with preeclampsia and 20 healthy controls. The preeclampsia group was subdivided into severe/non-severe and early/late-onset categories. Placental tissue samples underwent immunohistochemical analysis to assess LIF and VEGF expression in cytotrophoblasts, syncytiotrophoblasts, and villous stromal cells. Statistical comparisons and diagnostic accuracy parameters were calculated. RESULTS: LIF and VEGF expressions were significantly elevated in placental tissues from the preeclampsia group compared to controls. LIF levels increased notably in cytotrophoblast and syncytiotrophoblast cells. VEGF levels were higher in cytotrophoblasts, syncytiotrophoblasts, and villous stromal cells. When combined, syncytiotrophoblast LIF and VEGF levels yielded sensitivity of 77.5%, specificity of 95%, positive predictive value of 96.9%, and negative predictive value of 67.9% for predicting preeclampsia.

CONCLUSION: Elevated placental LIF and VEGF expressions are strongly associated with preeclampsia and correlate with disease severity. These factors hold significant potential as biomarkers, alone or combined, for early prediction of preeclampsia, facilitating timely interventions and improved clinical outcomes.

Keywords: LIF, VEGF, Placenta, Preeclampsia, Pregnancy, Trophoblast

Figure 1



Degree of VEGF and LIF immunohistochemical cytoplasmic staining in chorionic villi: No staining (A), Light staining (B), Moderate staining (C), Strong staining (D)

Degree of VEGF and LIF immunohistochemical cytoplasmic staining in decidua cells: No staining (A), Light staining (B), Moderate staining (C), Strong staining (D)

Immunohistochemical staining levels of placental cells with VEGF and LIF

Tablo 1

Table 1. Comparison of demographic and clinical parameters between preeclampsia and control groups

	Pre-eclampsia Group (n=40)	Control Group (n=20)	*p*
Age (years)	32.6 ± 4.0	31.9 ± 5.9	>0.05*
Gravida	3.0 ± 1.6	3.3 ± 2.1	>0.05
Parity	1.4 ± 1.2	1.5 ± 1.1	>0.05
BMI (kg/m2)	33.0 ± 7.0	29.3 ± 2.7	**<0.05**
OAB (mmHg)	118.7 ± 9.3	90.7 ± 3.9	**<0.05***
Right uterine artery pulsatility index	1.5 ± 0.7	1.0 ± 0.2	**<0.05**
Left uterine artery pulsatility index	1.3 ± 0.7	1.0 ± 0.2	>0.05
LIF decidual tissue cytoplasmic	5.9 ± 4.3	14.5 ± 6.8	**<0.05**
LIF syncytiotrophoblast cytoplasmic	69.0 ± 30.4	17.0 ± 7.9	**<0.05**
LIF cytotrophoblast cytoplasmic	74.1 ± 27.9	13.3 ± 5.8	**<0.05**
LIF villous stromal cytoplasmic	6.9 ± 4.7	11.5 ± 5.8	**<0.05**
LIF villous capillary cytoplasmic	3.9 ± 4.0	14.2 ± 8.7	**<0.05**
VEGF decidual tissue cytoplasmic	79.0 ± 29.2	67.4 ± 22.4	>0.05
VEGF syncytiotrophoblast cytoplasmic	126.1 ± 52.3	66.0 ± 19.4	**<0.05**
VEGF cytotrophoblast cytoplasmic	154.7 ± 48.4	73.2 ± 20.6	**<0.05**
VEGF villous stromal cytoplasmic	126.6 ± 62.2	45.8 ± 30.3	**<0.05**
VEGF villous capillary cytoplasmic	133.3 ± 93.3	93.2 ± 25.0	>0.05

BMI: Body mass index BMI: Mean arterial pressure
LIF: Leukocyte inhibitory factor VEGF: Vascular endothelial growth factor

*: Analyses were performed with Student's t-test.

Comparison of demographic and clinical parameters between preeclampsia and control groups

Tablo 2

Table 2. Comparison of preeclampsia and control groups with and without severe features in terms of LIF and VEGF

		Pre-eklampsia Group						P		
		Pre-eclampsia with severe features (n=7)		Pre-eclampsia without severe features (n=33)		Control Group (n=20)		P_1	P_2	P_3
LIF	Decidual tissue	5.7 ± 5.0		6.0 ± 4.2		14.5 ± 6.8		<0.05	<0.05	<0.05
	Syncytiotrophoblast	62.7 ± 38.8		76.3 ± 28.8		17.6 ± 7.9		<0.05	<0.05	<0.05
	Cytotrophoblast	75.4 ± 38.9		73.9 ± 26.3		13.3 ± 5.8		<0.05	<0.05	<0.05
	Villous stromal	7.0 ± 3.2		6.9 ± 5.2		11.5 ± 5.8		<0.05	<0.05	<0.05
	Villous capillaries	4.7 ± 2.9		3.7 ± 4.2		14.2 ± 8.7		<0.05	<0.05	<0.05
VEGF	Decidual tissue	92.7 ± 34.4		76.0 ± 27.7		67.4 ± 22.4		>0.05	>0.05	>0.05
	Syncytiotrophoblast	142.7 ± 39.9		122.8 ± 54.5		66.0 ± 19.4		<0.05	<0.05	<0.05
	Cytotrophoblast	177.4 ± 24.0		149.9 ± 51.1		73.2 ± 20.6		<0.05	<0.05	<0.05
	Villous stromal	149.4 ± 82.1		121.8 ± 57.8		45.8 ± 30.3		<0.05	<0.05	<0.05
	Villous capillaries	138.7 ± 38.5		132.2 ± 100		93.2 ± 25.0		>0.05	>0.05	>0.05

LIF: Leukocyte inhibitory factor VEGF: Vascular endothelial growth factor

P1: Comparison of preeclampsia group with severe features and preeclampsia group without severe features
P2: Comparison of preeclampsia group with severe features and control group
P3: Comparison of preeclampsia group without severe features and control group

Comparison of preeclampsia and control groups with and without severe features in terms of LIF and VEGF

Tablo 3

Table 3. Comparison of the subgroups of early-onset and late-onset preeclampsia according to severity characteristics with the control group in terms of LIF and VEGF

		Preeklampsia (n=40)					P							
		Early Onset		Late Onset										
		Preeclampsia with severe features (n=5)	Preeclampsia without severe features (n=17)	Preeclampsia with severe features (n=2)	Preeclampsia without severe features (n=16)	Control Group (n=20)	P1	P2	P3	P4	P5	P6	P7	P8
LIF	Decidual tissue	9.6±2.8	3.3±4.1	10.5±8.4	6.8±4.3	19.5±6.8	>0.05	>0.05	>0.05	>0.05	<0.05	<0.05	>0.05	<0.05
	Syncytiotrophoblast	46.3±27.8	71.3±32.1	104.5±10.6	87.4±31.2	17.6±7.9	>0.05	>0.05	>0.05	>0.05	>0.05	<0.05	<0.05	<0.05
	Cytotrophoblast	87.4±42.0	70.3±32.0	93.5±9.2	87.8±29.7	13.3±5.8	>0.05	>0.05	>0.05	>0.05	<0.05	<0.05	<0.05	<0.05
	Villous stromal	7.0±3.4	7.0±4.8	7.0±3.5	5.9±3.8	11.5±5.8	>0.05	>0.05	>0.05	>0.05	>0.05	>0.05	>0.05	>0.05
	Villous capillaries	4.4±3.6	3.8±3.2	5.1±4.2	3.1±2.9	14.2±8.7	>0.05	>0.05	>0.05	>0.05	<0.05	<0.05	>0.05	<0.05
VEGF	Decidual tissue	85.4±58.2	86.7±28.6	111.0±18.3	80.3±23.0	67.4±22.4	>0.05	>0.05	>0.05	>0.05	>0.05	>0.05	>0.05	>0.05
	Syncytiotrophoblast	140.6±47.5	135.7±47.7	153.0±18.3	121.4±61.5	66.0±19.4	>0.05	>0.05	>0.05	>0.05	<0.05	<0.05	>0.05	<0.05
	Cytotrophoblast	169.4±18.2	153.3±24.4	197.5±21.8	145.3±45.8	73.2±20.6	>0.05	>0.05	>0.05	>0.05	<0.05	<0.05	>0.05	<0.05
	Villous stromal	134.4±46.7	133.5±62.8	182.5±86.2	120.5±52.1	45.8±30.3	>0.05	>0.05	>0.05	>0.05	<0.05	<0.05	<0.05	<0.05
	Villous capillaries	113.4±68.7	141.3±108.9	103.0±31.4	120.6±81.2	93.2±25.0	>0.05	>0.05	>0.05	>0.05	>0.05	>0.05	>0.05	>0.05

LIF: Leukocyte inhibitory factor VEGF: Vascular endothelial growth factor

P1: Early onset severe pre-eclampsia subgroup compared with late onset severe pre-eclampsia subgroup
P2: Comparison of early-onset non-severe preeclampsia subgroup with late-onset non-severe preeclampsia subgroup
P3: Comparison of early-onset severe preeclampsia subgroup with late-onset non-severe preeclampsia subgroup
P4: Comparison of early-onset non-severe preeclampsia subgroup with late-onset severe preeclampsia subgroup
P5: Comparison of early onset severe pre-eclampsia subgroup with control group
P6: Comparison of early-onset preeclampsia subgroup without severe features and control group
P7: Comparison of late-onset severe pre-eclampsia subgroup with control group
P8: Comparison of late-onset preeclampsia subgroup without severe features and control group

Comparison of the subgroups of early-onset and late-onset preeclampsia according to severity characteristics with the control group in terms of LIF and VEGF

Tablo 4

Table 4. Correlation of LIF and VEGF levels in placental tissues with clinical and ultrasonographic characteristics of the patients

	MAP		Right uterine artery PI		Left uterine artery PI		BMI	
	r	p	r	p	r	p	r	p
LIF Desidual tissue	-0.546	**<0.01**	-0.359	**<0.01**	-0.246	>0.05	-0.021	>0.05
LIF Syncytiotrophoblast	0.679	**<0.01**	0.342	**<0.01**	0.321	**<0.05**	0.116	>0.05
LIF Cytotrophoblast	0.736	**<0.01**	0.270	**<0.05**	0.280	**<0.05**	0.183	>0.05
LIF Villous stromal	-0.225	>0.05	0.078	>0.05	0.041	>0.05	-0.143	>0.05
LIF Villous capillaries	-0.515	**<0.01**	-0.304	**<0.05**	-0.255	**<0.05**	-0.166	>0.05
VEGF Desidual tissue	0.188	>0.05	-0.073	>0.05	-0.168	>0.05	0.047	>0.05
VEGF Syncytiotrophoblast	0.603	**<0.01**	0.139	>0.05	0.096	>0.05	0.228	>0.05
VEGF Cytotrophoblast	0.620	**<0.01**	0.248	>0.05	0.218	>0.05	0.329	**<0.05**
VEGF Villous stromal	0.506	**<0.01**	0.003	>0.05	0.093	>0.05	0.262	**<0.05**
VEGF Villous capillaries	0.155	>0.05	-0.097	>0.05	-0.130	>0.05	-0.120	>0.05

LIF: Leukocyte inhibitory factor
VEGF: Vascular endothelial growth factor
MAP: Mean arterial pressure
PI: Pulsatility index
BMI Body mass index

Correlation of LIF and VEGF levels in placental tissues with clinical and ultrasonographic characteristics of the patients

Tablo 10

Variables	p	RR	%95 CI
LIF-DE	0.047	0.589	0.349-0.933*
LIF-VIST	0.394	0.909	0.731-1.131
LIF-VIKA	0.211	0.842	0.644-1.102
VEGF-DE	0.824	0.994	0.939-1.052
VEGF-VIST	0.055	1.051	0.999-1.107
VEGF-VIKA	0.551	1.006	0.986-1.226
BMI	0.562	1.084	0.826-1.422

Multiple logistic regression analysis in patients with LIF-DE (11 H Score), LIF-VIST (5.5 H Score), LIF-VIKA (5.5 H Score), VEGF-DE (77 H Score), VEGF-VIST (62.5 H Score) and VEGF-VIKA (105 H Score).

Tablo 5

	Sensitivity (%)	Spesifity (%)	PPV (%)	NPV (%)
LIF-DE	80	50	76.2	55.6
LIF-SIN	90	90	94.7	81.8
LIF-SIT	95	95	97.4	90.5
LIF-VIST	42.5	80	80.9	41
LIF-VIKA	75	75	85.7	60

Sensitivity, specificity, positive and negative predictive values of LIF-DE (11 H Score), LIF-SIN (27 H Score), LIF-SIT (22 H Score), LIF-VIST (5.5 H Score) and LIF-VICA (5.5 H Score) levels for the detection of pre-eclampsia when used as single parameters.

Tablo 6

	Sensitivity (%)	Spesifity (%)	PPV (%)	NPV (%)
LIF-SIN & VEGF-SIN	77.5	95	96.9	67.9
LIF-SIT & VEGF-SIT	82.5	100	100	74.1

Sensitivity, specificity, positive and negative predictive values for preeclampsia when LIF-SIN (27 H Score) and VEGF-SIN (78.5 H Score) and LIF-SIT (22 H Score) and VEGF-SIT (98.5 H Score) levels are used together.

Tablo 7

	Sensitivity (%)	Spesifity (%)	PPV (%)	NPV (%)
VEGF-DE	52.5	40	72.4	38.7
VEGF-SIN	82.5	90	94.3	72
VEGF-SIT	87.5	95	97.2	79.2
VEGF-VIST	80	80	88.9	66.7
VEGF-VIKA	60	75	82.8	48.4

Sensitivity, specificity, positive and negative predictive values of VEGF-DE (77 H Score), VEGF-SIN (78.5 H Score), VEGF-SIT (98.5 H Score), VEGF-VIST (62.5 H Score) and VEGF-VICA (105 H Score) levels in the detection of pre-eclampsia when used as single parameters.

Tablo 8

Variables	p	RR	%95 CI
LIF-DE	0.006	0.573	0.385-0.852*
LIF-VIST	0.227	0.899	0.756-1.069
VEGF-DE	0.526	0.986	0.943-1.031
VEGF-VIST	0.013	1.046	1.009-1.084*
BMI	0.493	1.096	0.858-1.375

Multiple logistic regression analysis in patients with LIF-DE (11 H Score), LIF-VIST (5.5 H Score), VEGF-DE (77 H Score) and VEGF-VIST (62.5 H Score).

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Retrospective evaluation of the antenatal corticosteroid effects on fetal thyroid functions in late preterm infants

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PURPOSE: This study aimed to evaluate the effects of maternal antenatal corticosteroid (ANS) administration on neonatal thyroid function in infants born between 34 and 37 weeks of gestation due to preterm labor.

MATERIALS-METHODS: A retrospective analysis was conducted using data from 99 pregnant women diagnosed with preterm labor and their infants, delivered between the 34th and 37th weeks of gestation at Karadeniz Technical University Hospital from 2015 to 2019. Participants were categorized into three groups: Group 1 included those who received a single dose (12 mg) of betamethasone, Group 2 received a double dose (24 mg in total), and Group 3 included those who did not receive ANS. Demographic characteristics, neonatal Thyroid Stimulating Hormone (TSH), and free Thyroxine (fT4) levels measured on the 5th postnatal day were compared among the groups.

RESULTS: There were no significant differences between groups in terms of maternal age, gestational age at delivery, or neonatal fT4 levels ($p > 0.05$). However, mean TSH levels were significantly lower in Group 2 (double-dose ANS) compared to Group 3 (no ANS) ($p < 0.05$). No significant difference in TSH or fT4 levels was observed between Groups 1 and 2, or between Groups 1 and 3 ($p > 0.05$). CONCLUSION: Double-dose antenatal corticosteroid therapy in late preterm pregnancies is associated with reduced neonatal TSH levels, suggesting a potential impact on early thyroid function. Further prospective studies are warranted to explore long-term clinical implications.

Keywords: steroid, neonatal, hypothyroidism, preterm, thyroid

Table 1: Descriptive statistics by ANS performance status groups

	Group 1 Single dose ANS n=21	Group 2 Double dose ANS n= 37	Group 3 ANS (-) n= 41
Maternal age (years)	30.7 ± 6.2 (21 – 42)	30 ± 5.1 (20 – 41)	30.8 ± 5.3 (19 – 39)
Gravida (median)	4 (1-8)	3 (1-8)	3 (1-9)
Parite(median)	2 (0-6)	1 (0-3)	2 (0-5)
Gestational week (week)	35 (34 – 36)	35 (34 – 36)	35 (34 – 36)
Neonatal birth weight (grams)	2603.1 ± 482.8 (1710 – 3400)	2530.3 ± 401.8 (1800 – 3500)	2821.2 ± 356.4 (2260 – 3600)
Umbilical cord pH	7.32 ± 0.04 (7.21-7.41)	7.35 ± 0.05 (7.26-7.43)	7.33 ± (7.18-7.41)
TSH, (mIU/L)	4.5 ± 2.2 (1.68 – 9.10)	3.8 ± 1.9 (1.24 – 8.92)	5.1 ± 3.4 (1.61 – 16.07)
fT4, (ng/dL)	1.56 ± 0.26 (0.99 – 2)	1.57 ± 0.26 (1.1 – 2.01)	1.55 ± 0.41 (0.80 – 2.44)

**Values are shown in the table as mean ± standard deviation (minimum – maximum).*

Table 2: Comparison of TSH and fT4 values of the all groups that received single dose ANS (group 1), received double dose ANS (group 2), the group that did not receive any ANS (group 3).

	Group 1 Single dose ANS n=21	Group 2 Double dose ANS n= 37	Group 3 ANS Not Done n= 41			
				p1	p2	p3
TSH, (mIU/L)	4.5 ± 2.2 (1.68 – 9.10)	3.8 ± 1.9 (1.24 – 8.92)	5.1 ± 3.4 (1.61 – 16.07)	0.250	0.399	0.042
fT4, (ng/dL)	1.56 ± 0.26 (0.99 – 2)	1.57 ± 0.26 (1.1 – 2.01)	1.55 ± 0.41 (0.80 – 2.44)	0.817	0.976	0.0797

** Values are shown in the table as mean ± standard deviation (minimum – maximum). p1:Comparison of TSH and fT4 values of the single dose ANS group and the double dose ANS group p2:Comparison of TSH and fT4 values of the group in which a single dose of ANS was administered and the group that did not receive ANS p3:Comparison of TSH and fT4 values of the group that received double dose ANS and the group that did not receive any ANS.*

SS-051

Fetal Type 2 Umbilical-Portal Venous Systemic Shunt: Prenatal Management and Outcomes of 7 Cases

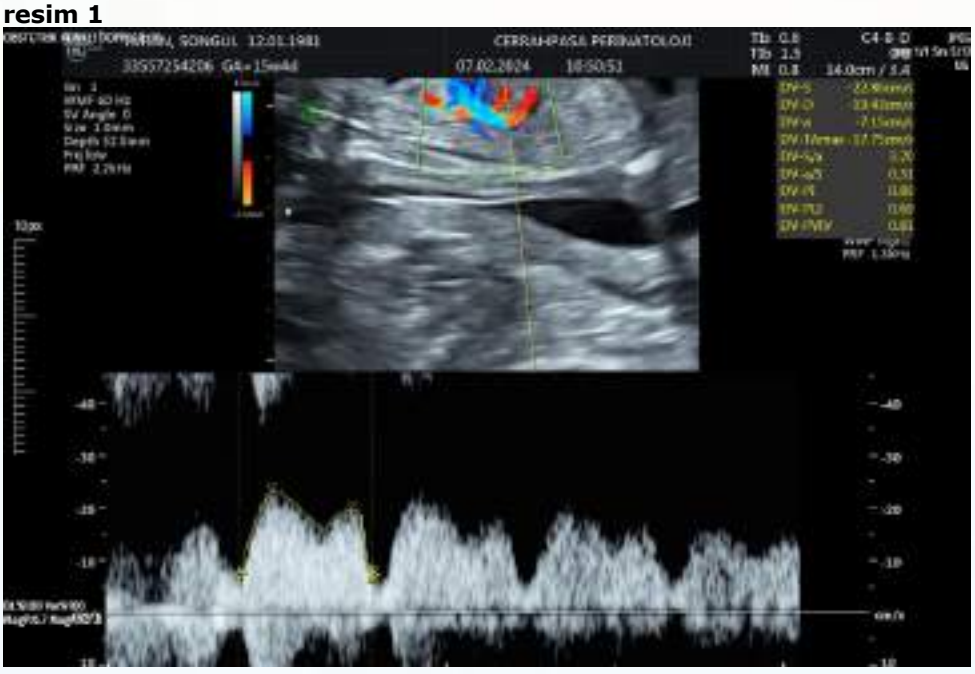
Dilek Buldum, ismail Yılmaz, Görkem Arıca, Aysegul Ozel, Ebru Alıcı Davutoğlu, Riza Madazli
İstanbul University Cerrahpasa, Cerrahpaşa Medical Faculty

Fetal Umbilical–Portal–Systemic Venous Shunts (UPSVS) are classified as vascular anomalies that present with a broad spectrum of clinical manifestations and prognoses. These shunts are anatomically classified into four types: Type I Umbilical–Systemic Shunt, Type II Ductus Venosus–Systemic Shunt, Type IIIa Intrahepatic Portal–Systemic Shunt, and Type IIIb Extrahepatic Portal–Systemic Shunt. This study presents seven fetuses that were diagnosed prenatally with Type 2 umbilicoportal systemic venous shunt. Every case was detected during routine second-trimester ultrasonography. To exclude chromosomal abnormalities, amniocentesis was performed in 4 patients. The other 3 patients chose not to accept the invasive procedures.In 2 cases that exhibited additional abnormalities (one had a hypoplastic nasal bone and the other was categorized as high risk in screening test), the amniocentesis results confirmed trisomy 21. These two pregnancies terminated with the consent of the families.In contrast, 2 cases that had no additional anomalies and presented with low-risk screening tests reached term and experienced no problems during their follow-up.The amniocentesis results for the remaining two cases have not been released.The study also noted a recent patient involving DCDA twin fetuses with selective fetal growth restriction where Type 2 UPSVS was detected in one fetus. An invasive procedure was recommended, and the decision is currently awaited.

Literature data has previously established the association of Type 2 upsvs with aneuploidy, specifically trisomy 21 (tri 21). Type 2 upsvs can be easily visualized during routine second-trimester ultrasounds. Therefore, patients must be informed about the potential risk of tri 21 when this finding is detected.

Keywords: umbilicoportal, shunt, ductus venous

resim 1



SS-052

Intrahepatic venous return abnormality in a fetus presenting with a cystic lesion in the first trimester: A case report

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Department of Obstetrics and Gynecology, Istanbul University-Cerrahpaşa Cerrahpaşa Medical Faculty, Istanbul, Turkey

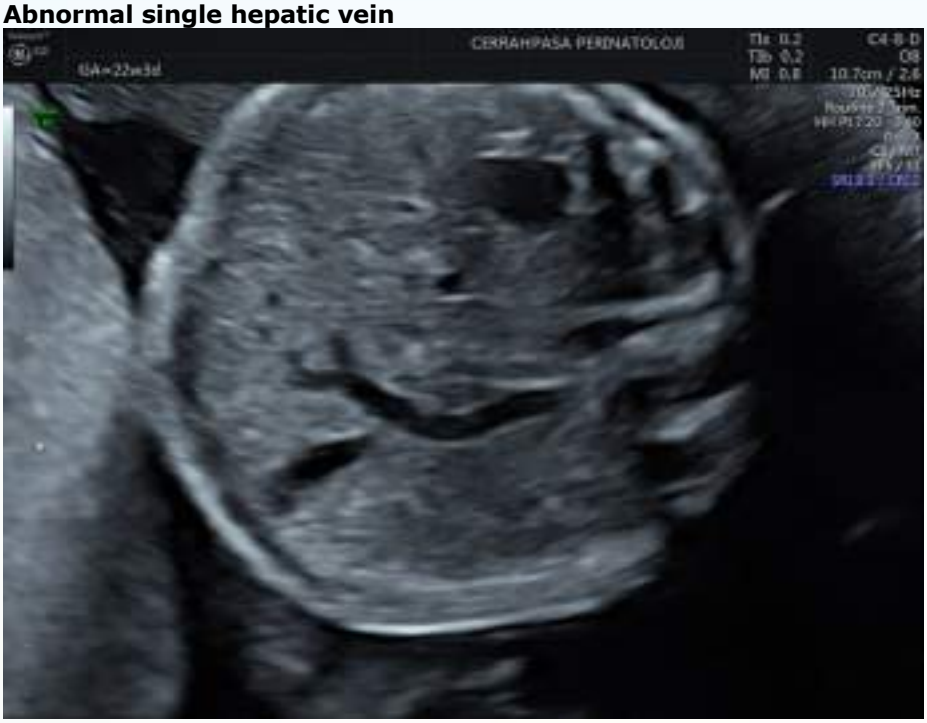
INTRODUCTION: The fetal liver has both afferent and efferent venous systems. The efferent system consists of the right, middle, and left hepatic veins draining into the inferior vena cava (IVC), while the afferent system includes the left portal vein (continuation of the umbilical vein), right portal vein, and main portal vein. Several abnormalities have been described, such as ductal agenesis and portosystemic shunts, but isolated efferent venous return abnormalities are extremely rare. We present a prenatally diagnosed case with postnatal confirmation and outcome.

CASE: A 27-year-old patient with a monochorionic diamniotic twin pregnancy conceived through IVF underwent first-trimester ultrasound, which revealed a cystic liver lesion in one fetus at 12 weeks. Amniocentesis at 15 weeks showed normal karyotype and microarray results. At 19 weeks, intrahepatic calcific lesions, a single umbilical artery, and abnormal venous return were detected. A single hepatic vein drained directly into the IVC, while the middle and left hepatic veins were absent. The portal system was otherwise normal. Fetal growth restriction did not develop. Delivery occurred by cesarean section at 35+6 weeks due to preterm labor. Postnatal imaging confirmed the findings, with additional right lobe hypoplasia and left lobe hyperplasia. The neonate is under pediatric surgical follow-up for hyperammonemia.

DISCUSSION: This case demonstrates that such anomalies can have metabolic consequences postnatally despite normal growth in utero. To the best of our knowledge, this is the first report of prenatal diagnosis, highlighting the value of systematic hepatic venous assessment during antenatal ultrasound.

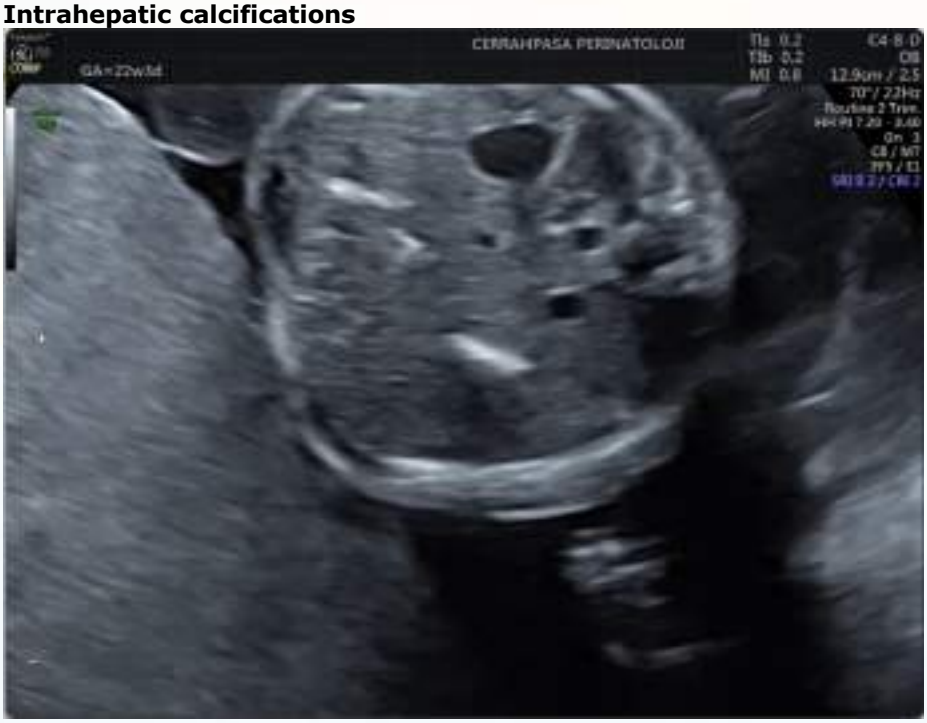
Keywords: Hepatic venous return abnormality, monochorionic monoamniotic twin pregnancy, prenatal diagnosis

Abnormal single hepatic vein



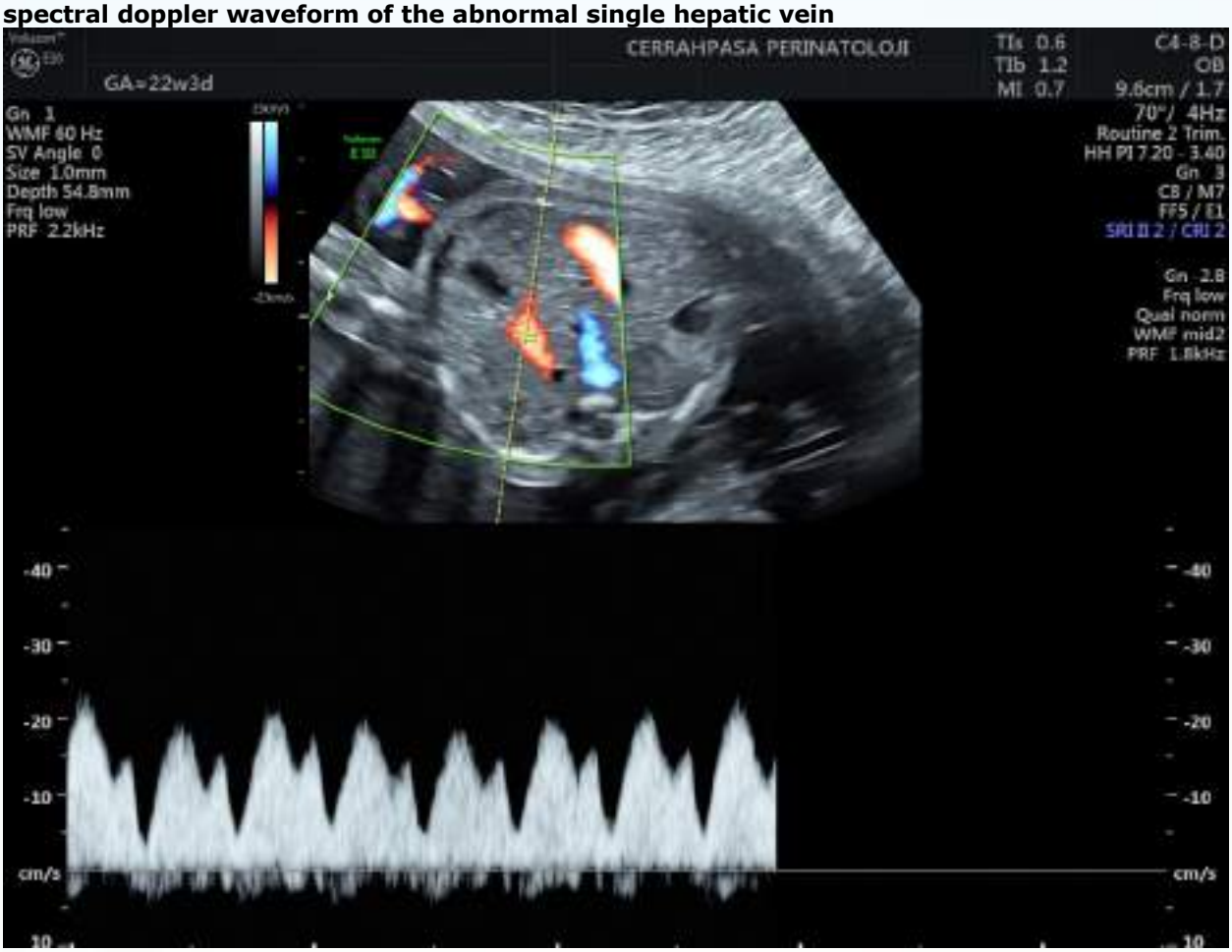
Liver efferent venous blood flow drained with abnormally located single hepatic vein

Intrahepatic calcifications



Intrahepatic calcific lesions

spectral doppler waveform of the abnormal single hepatic vein



typical venous doppler waveform of the abnormal single hepatic vein

Türkiye Maternal Fetal Tıp ve Perinatoloji Derneği
Ultrasonografi Kongresi
29 Ekim - 1 Kasım 2025
Renaissance Polat İstanbul Hotel - Yeşilköy

SS-053

Fetal Cardiac Myxoma Presenting with Increased Nuchal Translucency at 12 Weeks: The Earliest Reported Case in the Literature

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BACKGROUND: Fetal cardiac tumors are rare, with an estimated incidence of 0.14%–0.17%, and rhabdomyomas being the most frequently encountered type.Cardiac myxomas, although the most common benign cardiac tumors in adults, represent less than 1% of fetal cardiac tumors, making them an exceptionally rare prenatal diagnosis.While most fetal cardiac myxomas are diagnosed between 20 and 24 weeks of gestation, only a few have been identified as early as 13–14 weeks.To our knowledge, this is the earliest reported case of a prenatally diagnosed cardiac myxoma, identified at 12 weeks.

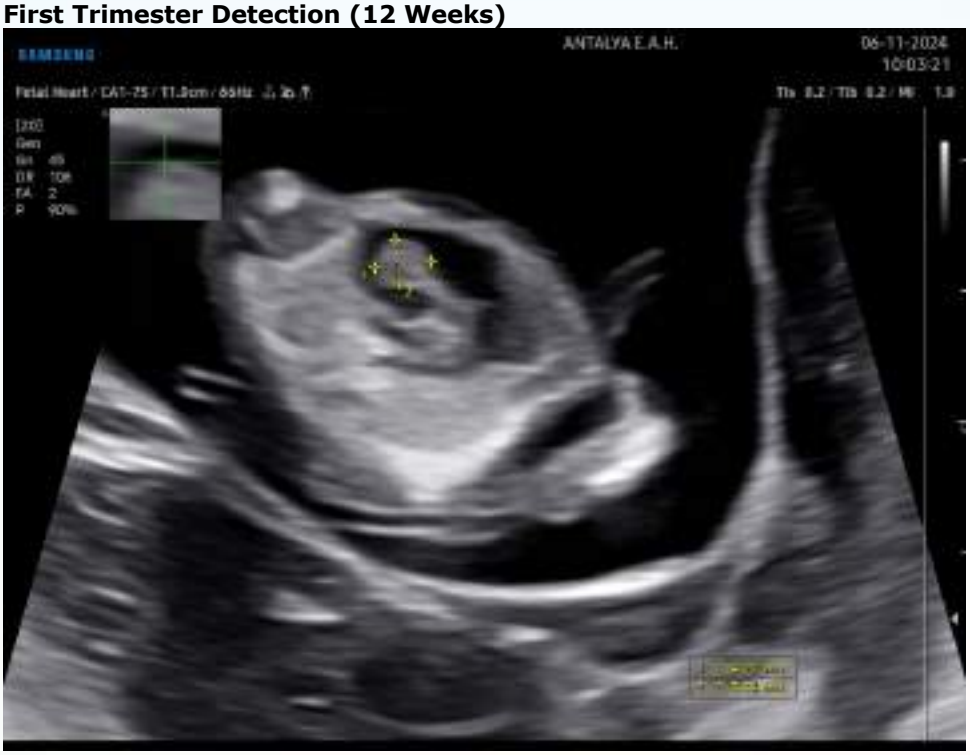
Case Presentation: A 37-year-old gravida was referred at 12 weeks due to increased nuchal translucency.First-trimester ultrasound revealed a 4×3 mm hyperechogenic, mobile mass in the right atrium, attached to the tricuspid valve, along with mild pericardial effusion.Chorionic villus sampling (CVS) was performed and yielded normal results.At 14 weeks, the mass had enlarged to 6×7 mm, with persistent pericardial effusion and reversed a-wave in ductus venosus.By 16 weeks, the lesion further obstructed right atrial inflow and showed ongoing hemodynamic compromise.Termination of pregnancy was elected due to the poor prognosis. Postmortem histopathology confirmed cardiac myxoma.

DISCUSSION: This is the earliest reported prenatal diagnosis of fetal cardiac myxoma, with progressive findings documented from the first trimester.Its solitary, mobile location on the tricuspid valve differentiated it from more common entities like rhabdomyoma or fibroma.Serial imaging was critical for clinical decision-making.

CONCLUSION: Although exceedingly rare, fetal cardiac myxomas can be detected as early as 12 weeks' gestation.Early diagnosis and close monitoring are essential for counseling and prognosis assessment.

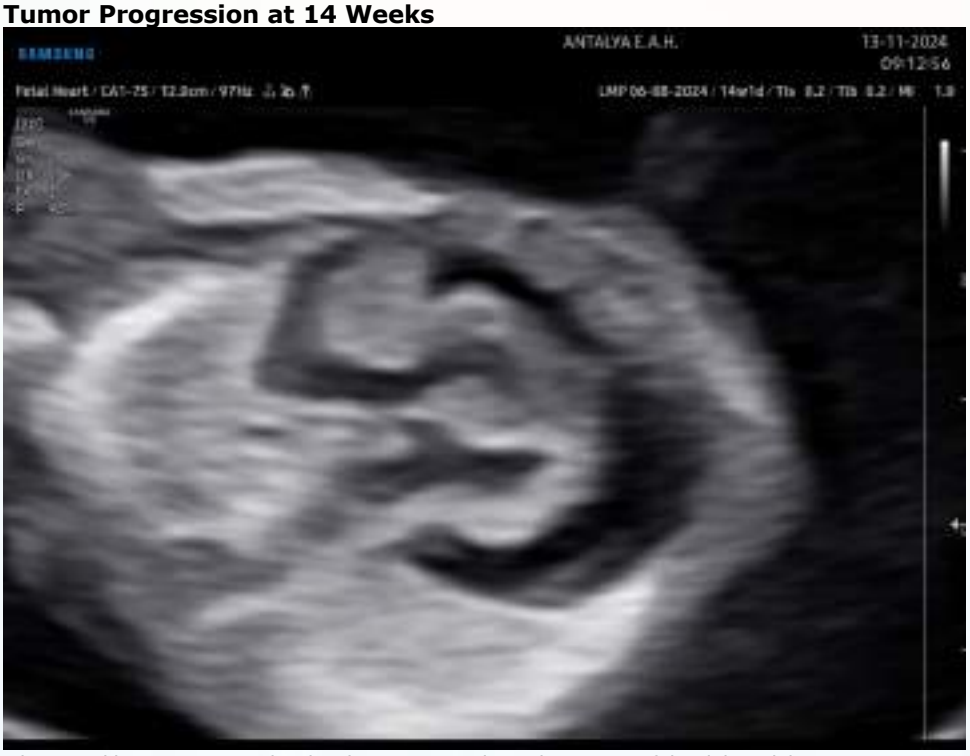
Keywords: Cardiac myxoma, Fetal cardiac tumor, First-trimester diagnosis, Prenatal ultrasound, Right atrium

First Trimester Detection (12 Weeks)



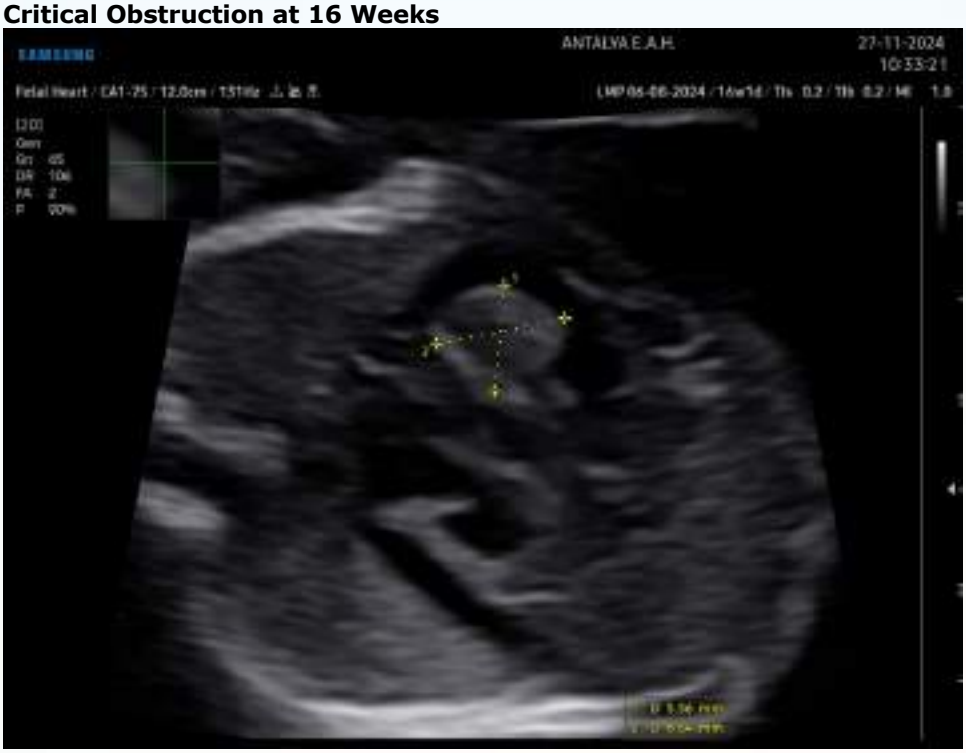
Fetal echocardiographic image obtained at 12 weeks' gestation demonstrating a 4×3 mm hyperechogenic, mobile mass within the right atrium, suggestive of cardiac myxoma.

Tumor Progression at 14 Weeks



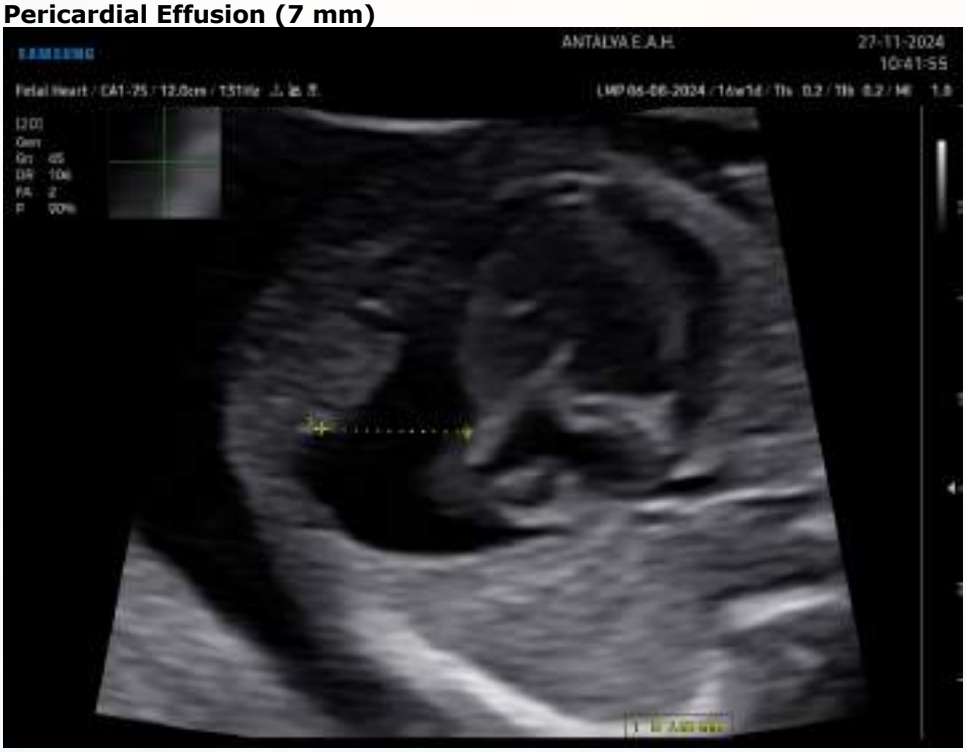
Ultrasound image at 14 weeks showing a progressive enlargement of the right atrial mass

Critical Obstruction at 16 Weeks



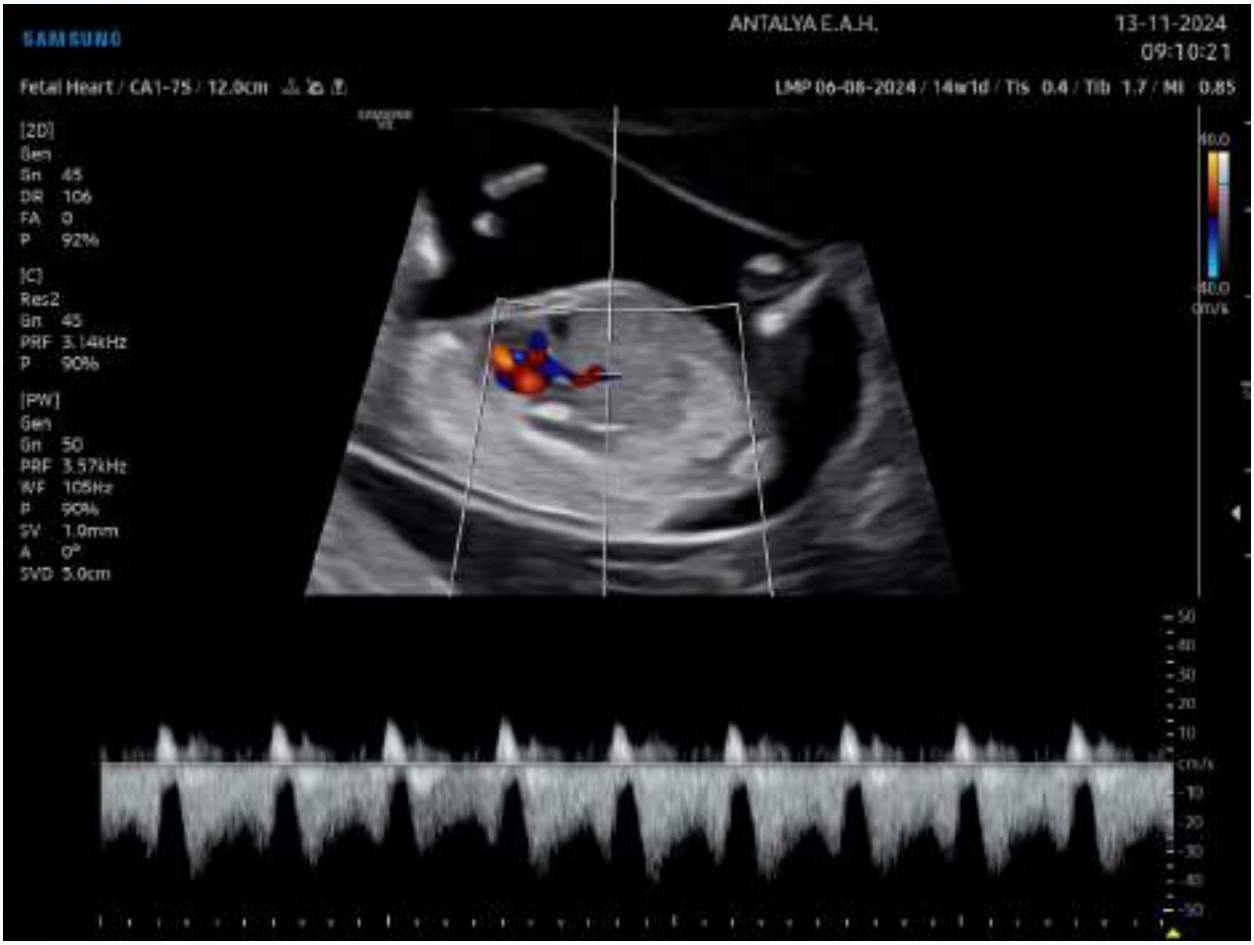
By 16 weeks, the tumor further enlarged to 5×6 mm, significantly impeding right atrial inflow and compressing the tricuspid valve.

Pericardial Effusion (7 mm)



Axial view showing pericardial effusion measuring approximately 7 mm surrounding the fetal heart.

Ductus Venosus with Reversed A-Wave



Doppler waveform of the ductus venosus demonstrating a reversed a-wave at 14 weeks, consistent with impaired cardiac compliance.

SS-054

Prenatal Findings and Postnatal Course of Fetuses Diagnosed With Cardiac Tumors

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INTRODUCTION: Cardiac tumors are rare anomalies, with an incidence ranging from 0.17 to 28 per 10,000 births, mostly reported in autopsy studies. Prenatal diagnosis plays a critical role in pregnancy management and postnatal outcomes[1]. This case series presents prenatal and postnatal findings of four fetuses diagnosed with cardiac tumors at Başakşehir Çam and Sakura City Hospital.

METHODS: Four fetuses diagnosed with cardiac tumors between 2020-2024 were retrospectively evaluated. Gestational week at diagnosis, tumor location, associated anomalies, delivery mode, and postnatal prognosis were analyzed.

RESULTS: In Case 1, a 35×40 mm cystic, hyperechogenic mass was detected in the posterior right atrium.In Case 2, a 25×32 mm homogeneous, solid, hyperechogenic mass occupied the entire left ventricle. Case 3 had an 8×6 mm echogenic mass in the interventricular septum extending into the myocardium, consistent with rhabdomyoma. In Case 4, a solitary hyperechogenic mass extended from the left ventricle into the posterior left atrium. Only Case 3 underwent amniocentesis, which was negative for tuberous sclerosis and other genetic disorders. All fetuses had pericardial effusion and were delivered via cesarean section at term.All newborns survived; one underwent successful surgical resection. Postnatal diagnoses included rhabdomyoma, extracardiac teratoma, fibroma and lipoma.

Discussion & CONCLUSION: Rhabdomyomas are generally benign and often associated with tuberous sclerosis[2]. Teratomas are more aggressive and commonly linked to hydrops[2]. In this series, only the teratoma case required surgery. No postnatal mortality occurred. This case series emphasizes the importance of prenatal diagnosis and multidisciplinary management in optimizing outcomes for different fetal cardiac tumor types.

Keywords: cardiac tumor, rhabdomyoma, tuberosclerosis

SS-055

Fetal Arrhythmias: Diagnosis and Management of Supraventricular Tachycardia – Two Case Reports

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Ordu Education and Research hospital

INTRODUCTION: Fetal tachycardia, defined as a sustained ventricular rate >180 bpm, may cause intrauterine morbidity and mortality. Supraventricular tachycardia (SVT) is the most common type, accounting for 66–90% of cases. Early recognition and therapy improve outcomes. We present two non-hydropic SVT cases and discuss management.

Case 1: A 29-year-old primigravida was diagnosed with sustained tachycardia (220 bpm) at mid-trimester scan. Spectral Doppler confirmed 1:1 AV conduction with AV < VA intervals. Pericardial effusion was present. She was hospitalized and treated with digoxin (2×0.5 mg) and flecainide (2×100 mg). Within 48 hours, rhythm normalized and effusion resolved. At 39 weeks, she delivered a 3250 g infant. Postnatal evaluation was normal.

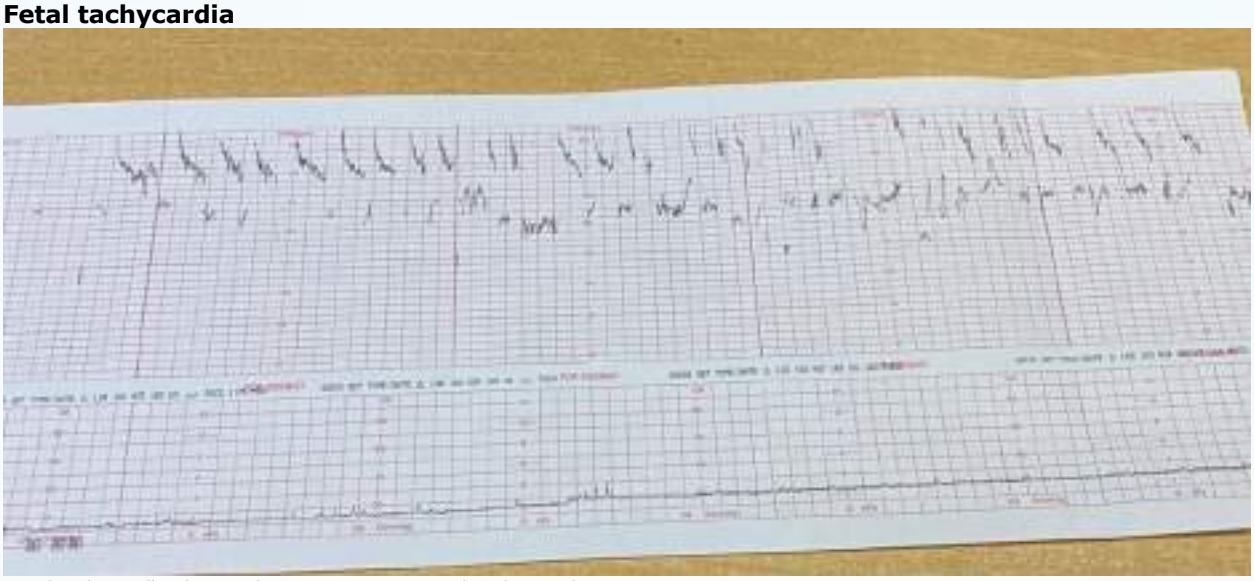
Case 2: A 30-year-old primigravida was referred at 34+4 weeks for tachycardia (200-220 bpm). Spectral Doppler showed normal AV relation with short VA interval. No effusion was seen. Digoxin (2×0.25 mg) and flecainide (2×100 mg) restored rhythm within 24 hours. At 36+4 weeks, tachycardia recurred; digoxin was increased to 0.5 mg with control achieved. Stable at 37+4 weeks.

Discussion SVT is the most frequent fetal tachyarrhythmia and may progress to hydrops. Diagnosis relies on spectral Doppler assessment of AV and VA intervals. The goal is to prevent deterioration and prolong pregnancy to term. Preterm delivery is not advised unless instability occurs. Digoxin is the preferred first-line drug; flecainide or sotalol may be added in resistant or hydropic cases. In both patients, combined digoxin and flecainide achieved control.

CONCLUSION: Early diagnosis, close surveillance and multidisciplinary care enable favorable outcomes and term delivery.

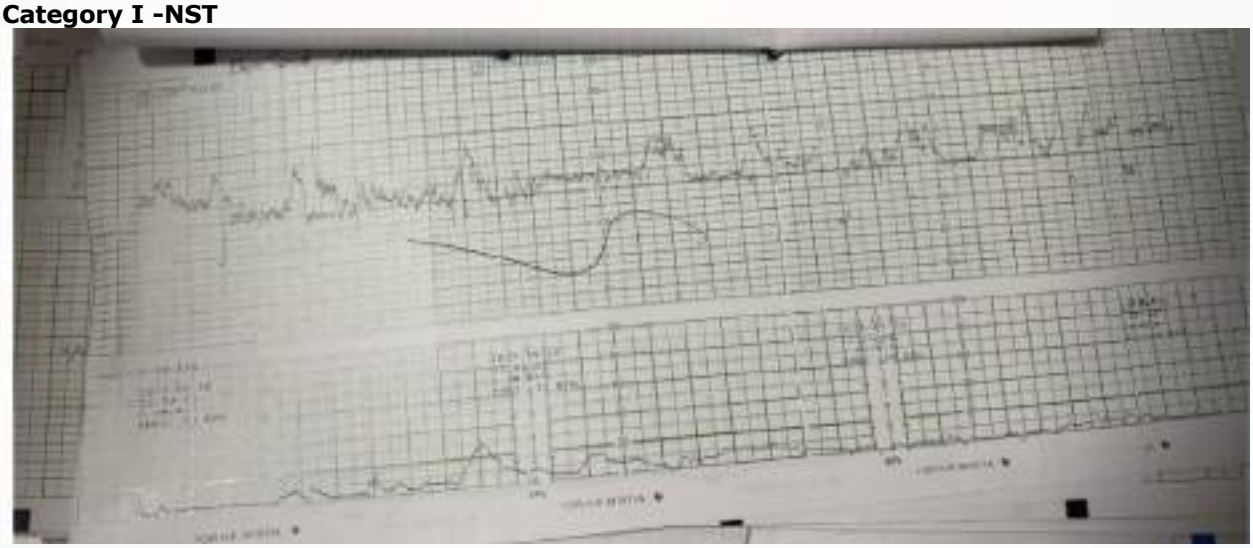
Keywords: Antiarrhythmic therapy, Fetal arrhythmia, Spectral Doppler

Fetal tachycardia



Fetal tachycardia observed on NST at 34+4 weeks of gestation.

Category I -NST



A Category I NST was recorded within 24 hours following the administration of antiarrhythmic therapy, demonstrating normal baseline, moderate variability and absence of pathological decelerations.

SS-056

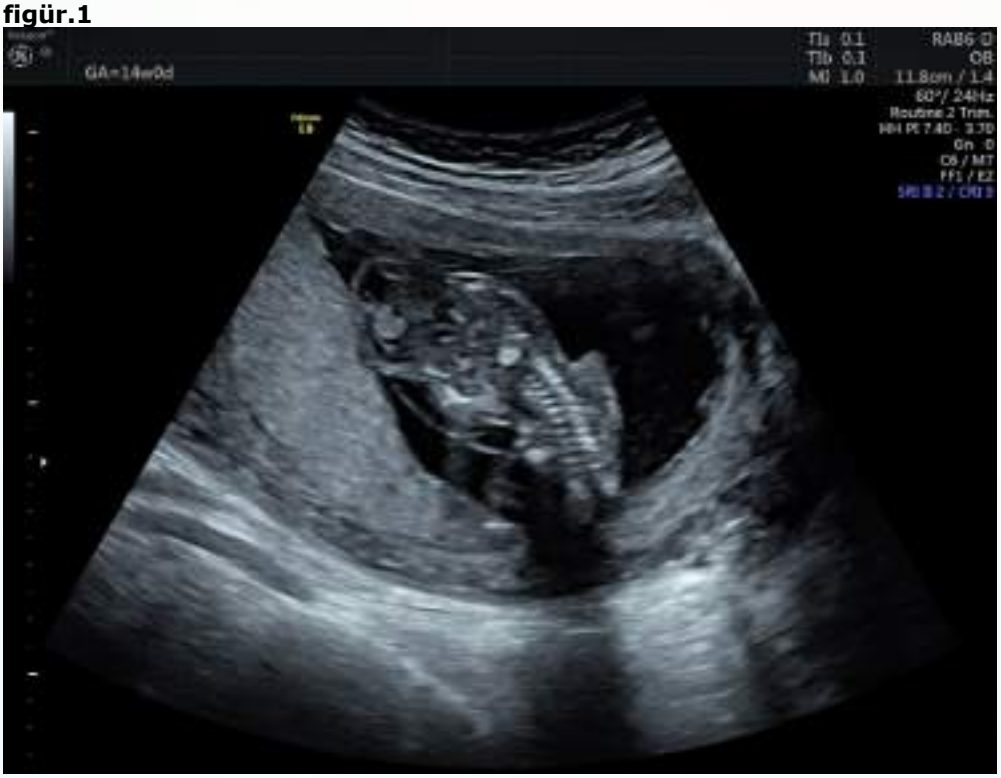
Cantrell Pentalojisine Eşlik Eden Amelia Olgu Sunumu

Engin Öztürk, GULSAH AYNAOGLU YILDIZ
Ankara Üniversitesi Tıp Fakültesi, Perinatoloji Bilim Dalı, Ankara

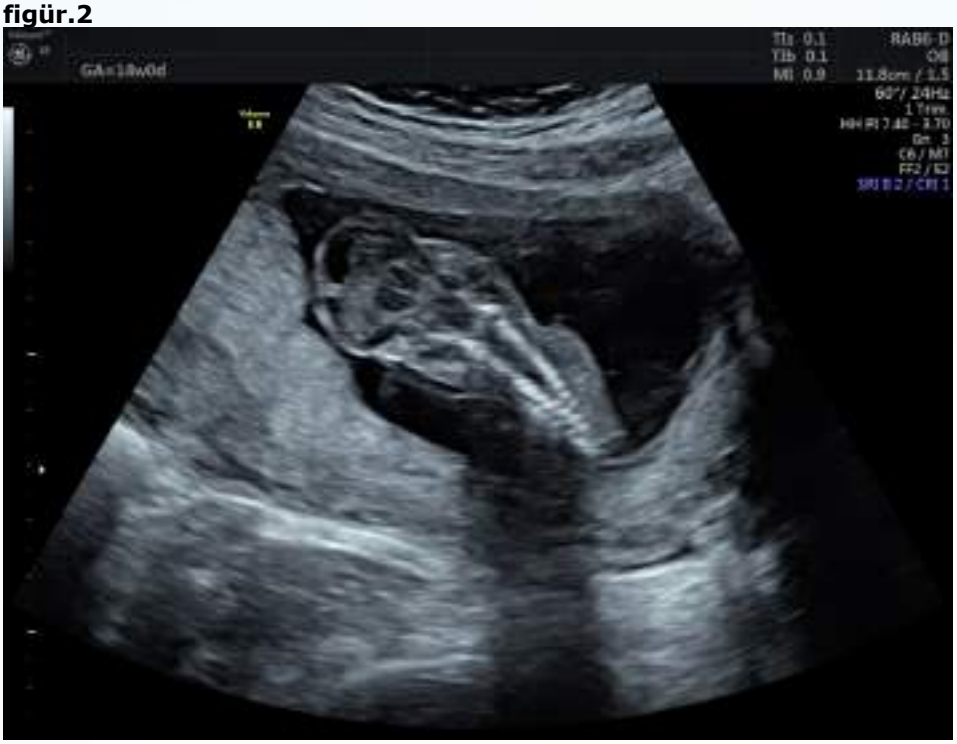
Cantrell pentalojisi nadir görülen bir doğumsal anomalidir; prevalansı 1/65.000-1/200.000 arasında değişmektedir. Bu sendrom, ilk olarak 1958 yılında, orta hatta supraumblikal torakoabdominal duvar defekti, anterior ve perikardiyal diyafram defekti, sternum alt bölüm defekti ve kalp anomalisi belirlenen olgularda Cantrell ve arkadaşları tarafından tanımlanmıştır. Cantrell pentalojisi tanısı koymak için yukarıda sayılan beş ölçütün de bulunması gerekir. Cantrell pentalojisinde genetik olarak Xq25-q26.1 ve X-linked PORCN mutasyonları bulunmaktadır. Olgu 14 haftalıkken tarafımıza ikili tarama için başvurmuş olup cantrel pentalojisine eşlik eden amelia bulgusu saptanmıştır. Aynı zamanda sonografik olarak ensefalosel, omfalosel, ektopia kordis, pes eqinavarus, tek umblikal arter, vertebral anomali, yarık damak ve dudak izlenmiştir. Hastaya genetik incelemeye yönelik olarak vajinal cvs yapılmış ve kromozomal patoloji izlenmemiştir. Terminasyon planlanmış ve hasta termine edilmiştir. Cantrell pentalojisi nadir görülen ve postnatal dönemde mortalite oranı yüksek yapısal defektler içeren bir sendromdur. Erken prenatal dönemde tanısının konulması ve aileye bu sendromun prognozu, etiyolojisininde kromozomal mutasyonların olabileceği anlatılmalıdır. Mutlaka genetik inceleme yapılması konusunda bilgilendirilerek gebeliğin erken dönemde terminasyonu sağlanmalıdır.

Anahtar Kelimeler: Amelia, Cantrell pentalojisi, Vajinal koryonik villüs örnekleme

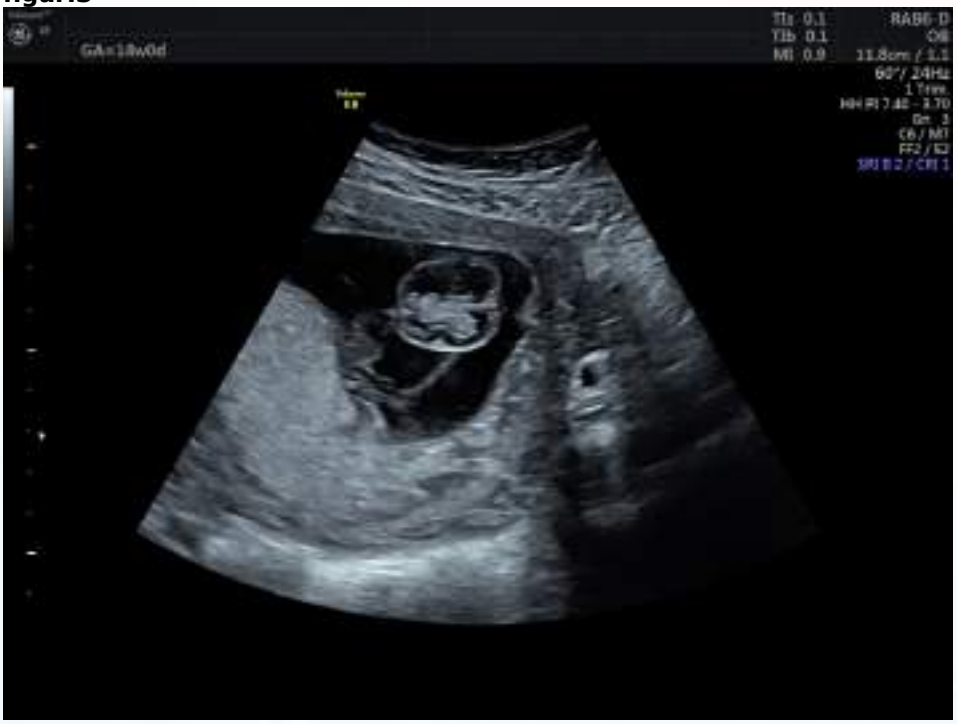
figür.1



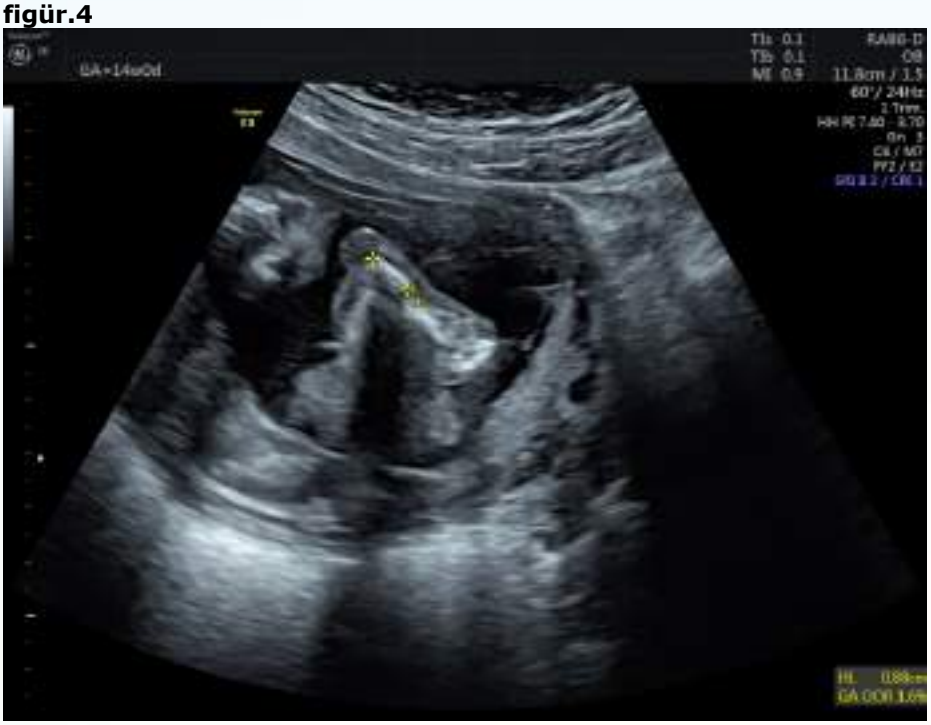
figür.2



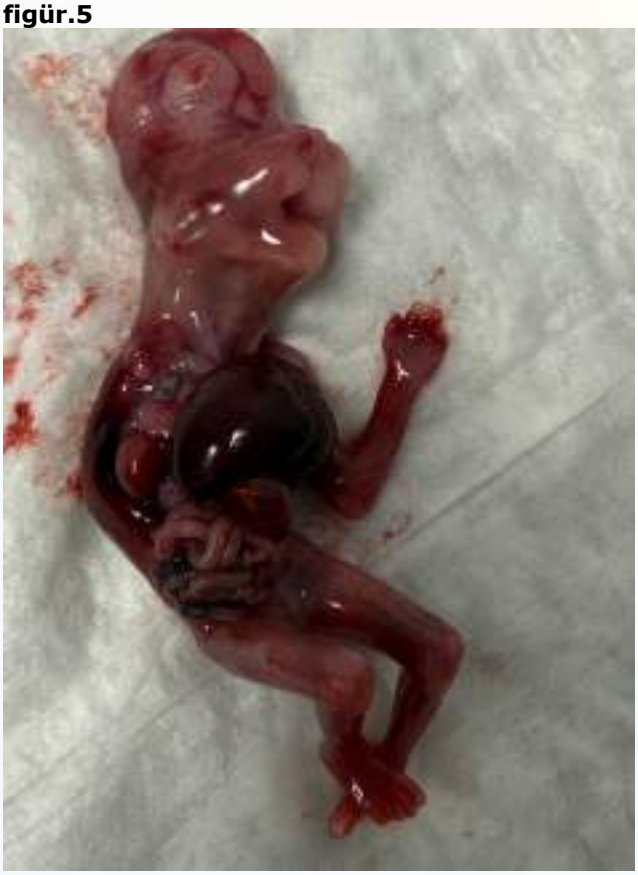
figür.3



figür.4



figür.5



SS-057

Prenatal Diagnosis Of Ebstein's Anomaly By Ultrasonography: A Case Report

Begüm Aktaş Çiftçi, Turgay Hakkı Emet, Ayşegül Ünal Gören, Kıvılcım Gören, Tahir Kaçar, Merve Baştan, Selçuk Özden
Sakarya Üniversitesi Eğitim Ve Araştırma Hastanesi, Perinatoloji Kliniği, Sakarya, Türkiye

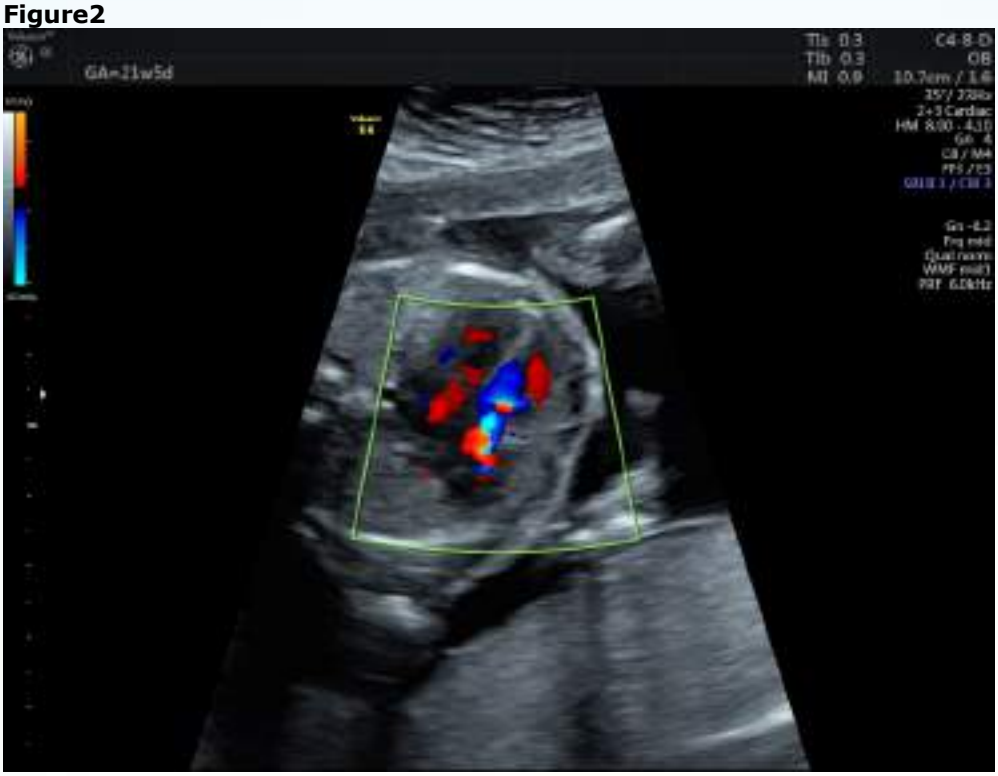
OBJECTIVE: Ebstein's anomaly is a rare and complex malformation, accounting for approximately 1% of congenital heart diseases(1). It is characterized by the downward displacement of the tricuspid valve leaflets, atrialized appearance of the right ventricle, and tricuspid regurgitation(2). In this study, we present a case diagnosed with Ebstein's anomaly using prenatal ultrasonography and fetal echocardiography, emphasizing the importance of early prenatal diagnosis.

CASE: A 33-year-old pregnant woman, G4P1A2, presented for the first time at 12+4 weeks of gestation. Ultrasonography revealed increased nuchal translucency (NT: 3.5 mm) and cystic hygroma; however, the patient declined genetic testing. In the following weeks, right atrial dilatation, tricuspid valve abnormality and regurgitation, and pulmonary artery stenosis were observed. In subsequent follow-ups, the characteristic findings of the anomaly became more evident.

CONCLUSION: As reported in the literature(3), Ebstein's anomaly may not always present with clear findings in early pregnancy. However, with careful follow-up and fetal echocardiography, the classic sonographic features can be revealed, allowing diagnosis.

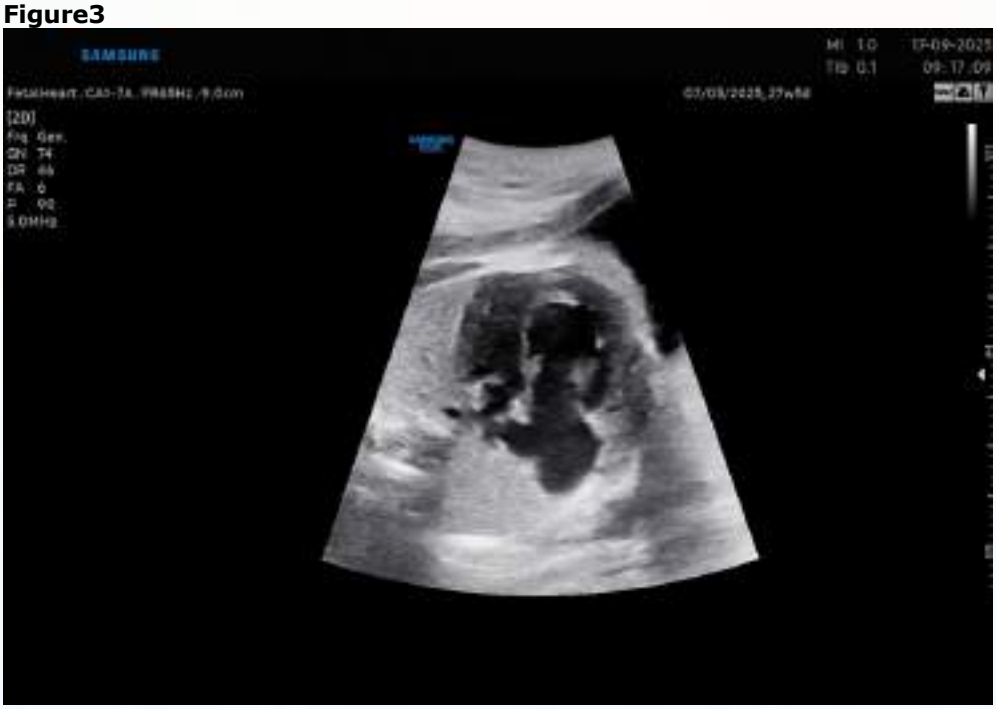
Keywords: Ebstein's anomaly, fetal echocardiography, tricuspid regurgitation

Figure2



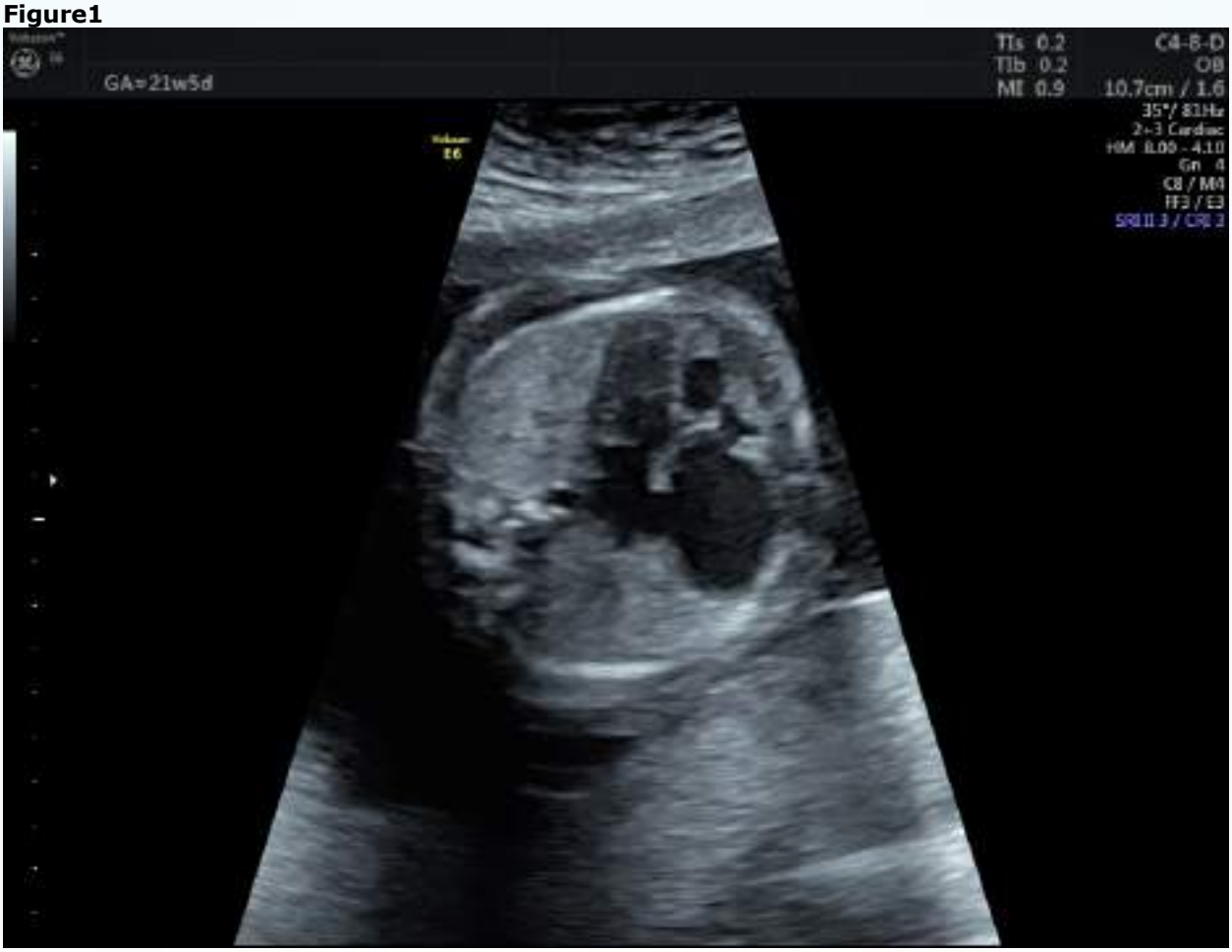
tricuspid regurgitation in the fetal heart

Figure3



Downward displacement and lengthening of the septum and anterior valves on fetal heart ultrasound

Figure1



Right atrial dilatation in the fetal heart

SS-058

Case Presentation: Homozygous Mutation in the Protein Tyrosine Kinase 7 (PTK7) Gene

Esin Şahin Toruk, Pınar Tokdemir Çalış, Hatice Aydın Salman, Arzu Sümer Ortaç, Esra Nuhoğlu Bilgin, Bilal Emre Erzeneoğlu, Deniz Karçaaltıncaba
Division of Perinatology, Department of Obstetrics and Gynecology, Gazi University Faculty of Medicine, Ankara, Turkey

BACKGROUND: Neural tube defects are multifactorial congenital anomalies resulting from the failure of neural tube closure during early embryogenesis. In addition to environmental factors, genetic determinants such as disruptions in the planar cell polarity signaling pathway have been implicated in their pathogenesis. The Protein Tyrosine Kinase 7 (PTK7) gene encodes a transmembrane protein involved in PCP signaling, and animal models have demonstrated its role in neural tube closure and organ morphogenesis. However, the phenotypic spectrum of PTK7 mutations in humans remains poorly defined.

Case Presentation: We report a 24-year-old gravida 3 para 2 living 1 (G3P2L1). Her previous pregnancy was terminated at 23 weeks due to meningomyelocele. In the current pregnancy, prenatal ultrasonography at 17 weeks revealed lumbosacral open spina bifida, bilateral hyperechogenic cystic kidneys with pyelectasis, and right rocker-bottom foot. Serial scans identified additional anomalies including bilateral ventriculomegaly, vermian agenesis, cerebellar "banana sign," umbilical vein varix, undescended testes, and hypospadias. At 37 weeks, she delivered a male infant weighing 2830 g with Apgar scores of 8 and 9. Postnatal genetic testing identified a homozygous PTK7 mutation.

CONCLUSION: This case illustrates a rare PTK7-related multisystem phenotype involving the central nervous system, kidneys, genitourinary tract and skeletal system. The findings support the role of PTK7 not only in neural tube closure but also in broader embryonic morphogenesis. Given the consanguinity and recurrence in a prior pregnancy, genetic counseling and targeted prenatal testing are recommended in at-risk families.

Keywords: Neural tube defect, PTK7, planar cell polarity, spina bifida, congenital anomalies, genetic counseling

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Ultrasonografi Kongresi
29 Ekim - 1 Kasım 2025
Renaissance Polat İstanbul Hotel - Yeşilköy

SS-059

Prenatal diagnosis and clinical management of multicystic encephalomalacia: A Case Report

Mehtap Korkut[1], Bilge Kapudere[2], Zehra Tavukçuoğlu[2], Reyhan Ayaz[2], Elif Yilmaz Gulec[3]
[1]Department of Obstetrics and Gynecology,Istanbul Medeniyet University,Istanbul,Turkey
[2]Department of Perinatology,Istanbul Medeniyet University,Istanbul,Turkey
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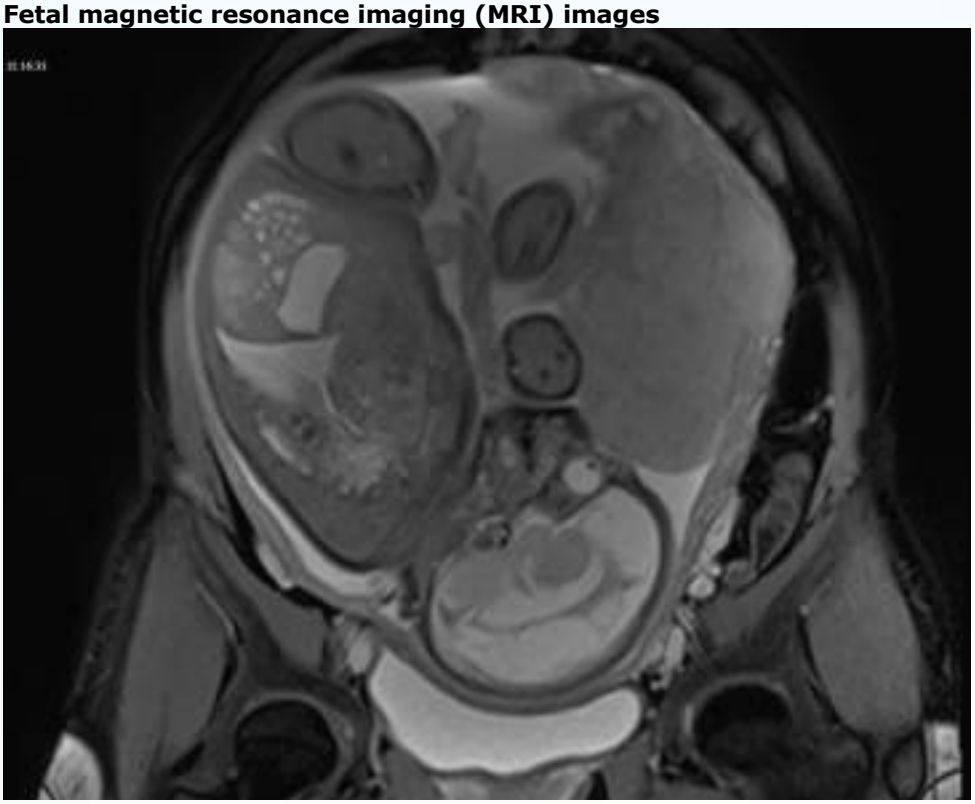
BACKGROUND: Multicystic encephalomalacia(MCE) is a rare encephalopathic disorder characterized by multiple thin-walled cystic cavities within the cerebral parenchyma, most commonly resulting from hypoxic-ischemic brain injury.It typically manifests in the late third trimester and is associated with unfavorable neurological outcomes.

CASE: We present the case of a 28-year-old multiparous woman whose fetus was diagnosed prenatally with multicystic encephalomalacia at 38+1 weeks of gestation.The patient was referred for evaluation due to ventriculomegaly.Ultrasonography demonstrated dilatation of the posterior horns of the lateral ventricles measuring 12 mm, while the cerebral parenchyma could not be clearly assessed.A hypoechoic cystic lesion was also observed within the cerebral region. The diagnosis was subsequently confirmed by fetal magnetic resonance imaging(MRI).After comprehensive counseling regarding the poor prognosis, including the risk of severe neurodevelopmental sequelae such as profound psychomotor retardation, epilepsy, and dystonia, the patient elected pregnancy termination. Extensive infectious and genetic evaluations were performed. Amniocentesis and cordocentesis were performed for CMV PCR, Toxoplasma, ParvovirusB19, and Rubella testing, as well as for karyotyping, chromosomal microarray analysis, and clinical exome sequencing. Intracardiac potassium chloride was administered to induce fetal demise.Exome analysis revealed potential associations between the fetal findings and variants in RNF213(Moyamoya disease) and MOCS2(molybdenum cofactor deficiency).

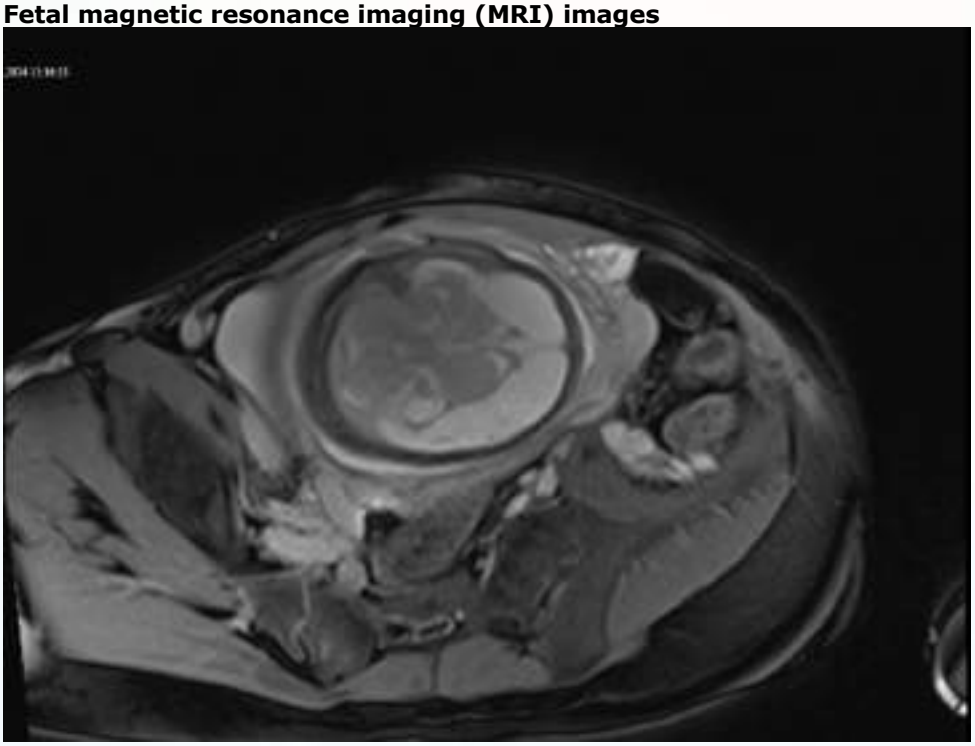
CONCLUSION: Prenatal diagnosis of multicystic encephalomalacia carries a poor prognosis with a high likelihood of severe neurological impairment or perinatal mortality. Early recognition is essential for accurate parental counseling and informed decision-making regarding management options, including the possibility of pregnancy

Keywords: Multicystic encephalomalacia, fetal MRI, prenatal diagnosis

Fetal magnetic resonance imaging (MRI) images



Fetal magnetic resonance imaging (MRI) images



Fetal Ultrasound Images Demonstrating Multicystic Encephalomalacia

Fetal Ultrasound Images Demonstrating Multicystic Encephalomalacia

SS-060

A rare case report of MCKUSICK-KAUFMAN syndrome

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Background: McKusick-Kaufman syndrome (MKKS) is a rare autosomal recessive disorder characterized by the triad of hydrometrocolpos, postaxial polydactyly, and congenital heart disease. Prenatal diagnosis is challenging due to variable sonographic findings. Early recognition is important for parental counseling and postnatal management.

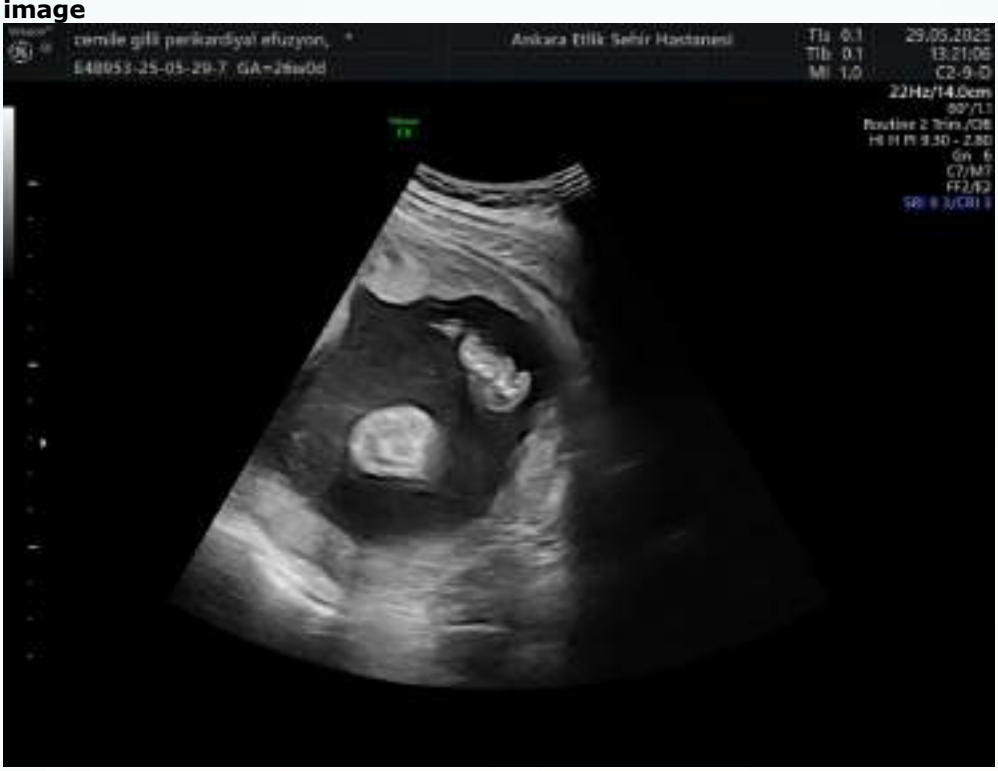
Case: A 19-year-old primigravida at 23+2 weeks underwent fetal anatomical evaluation without prior screening. Ultrasound revealed bilateral enlarged echogenic dysplastic kidneys, polydactyly of the left hand and foot, and a cystic pelvic mass posterior to the bladder consistent with hydrometrocolpos. Additional findings included minimal ascites, pericardial effusion, aortic arch hypoplasia, and inlet ventricular septal defect. Amniocentesis, fetal MRI, and pediatric cardiology consultation were recommended but declined. The findings were suggestive of MKKS.

At 39+5 weeks, cesarean section was performed for fetal distress. A female neonate weighing 3695 g with APGAR scores of 7/9 was delivered. Postnatal ultrasound showed multiple renal cysts (<7 mm) consistent with dysplasia. Clinical features included imperforate hymen with a pelvic mass (~10 cm) drained bedside and congenital cardiac anomalies. Echocardiography confirmed transitional atrioventricular septal defect with aortic valve and arch hypoplasia. Genetic analysis is pending.

CONCLUSION: This case highlights the association of renal dysplasia, urogenital malformations, and complex cardiac anomalies in MKKS. Comprehensive prenatal evaluation and multidisciplinary neonatal care are essential for prognosis and parental counseling.

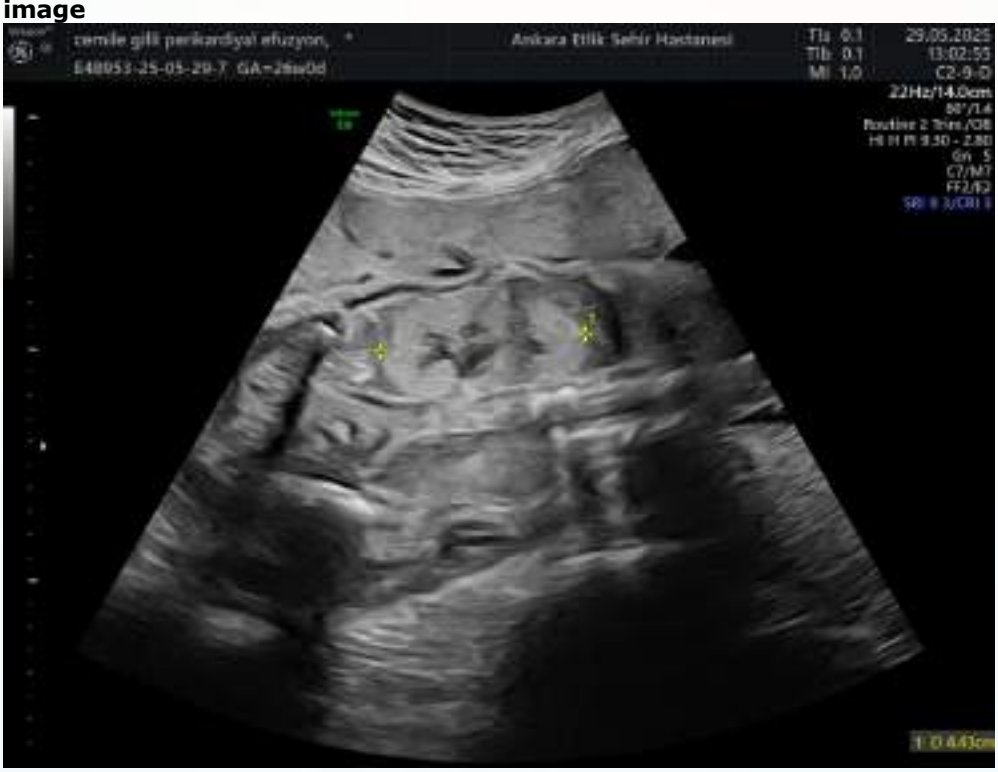
Keywords: McKusick-Kaufman syndrome, renal dysplasia, hydrometrocolpos, prenatal diagnosis

image



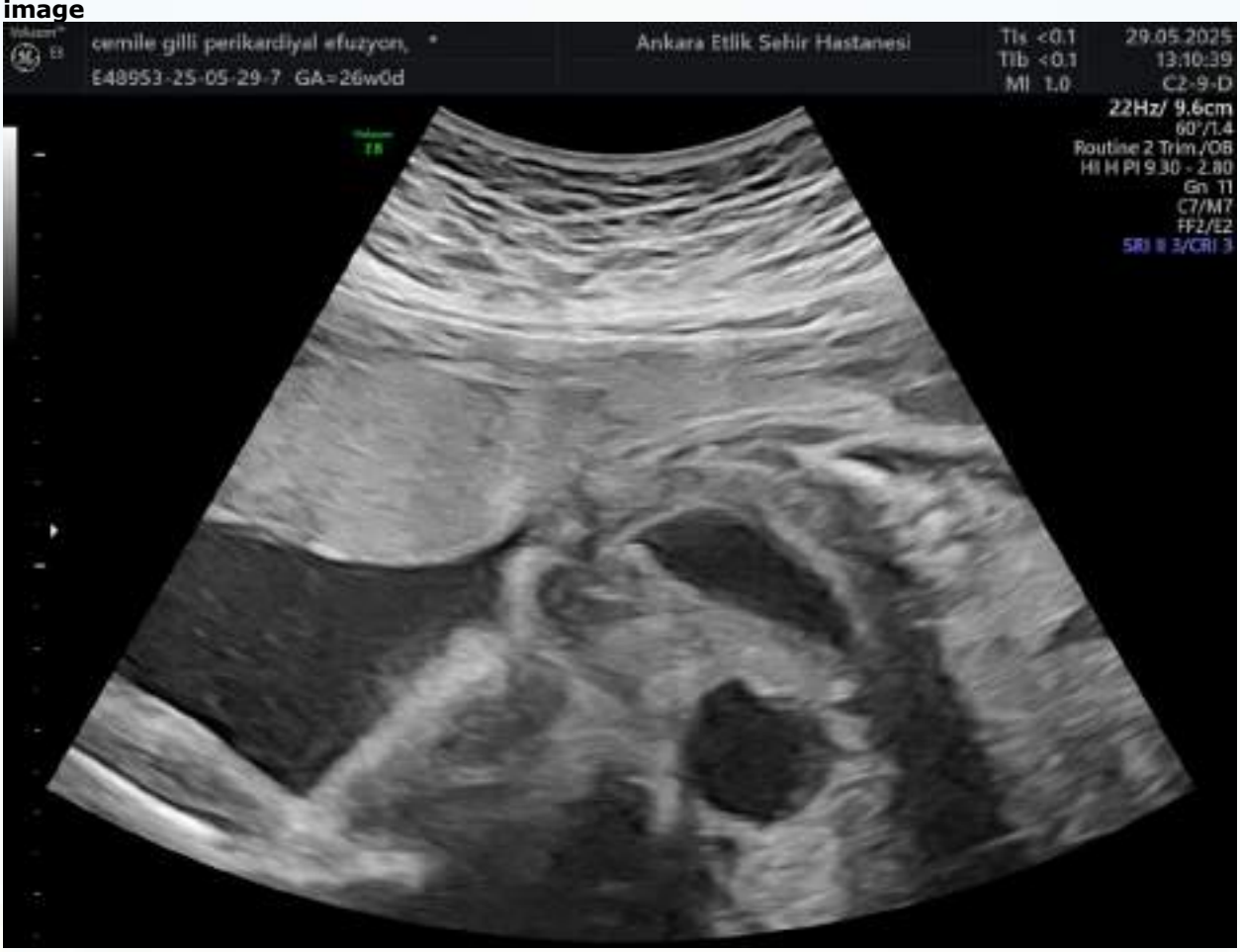
polydactyly

image



HYPERECHOGENIC KIDNEY

image



BLADDER AND HYDROCOLPOS IMAGE

SS-061

Case report: a hydrops fetalis case associated with a PIEZO1 gene mutation

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Fetal pleural effusion is defined as a nonspecific fluid collection located in the pleural space and can occur as a primary or secondary effusion. The estimated incidence of hydrothorax has been reported as 1: 10,000 to 1: 15,000 pregnancies with male predominans. Aneuploidy found in 6%–17% of affected pregnancies. Neonatal prognosis of hydrothoraces varies from spontaneous prenatal resolution (22%) to generalized hydrops fetalis or even death. Prenatal therapy including drainage and shunting of the pleural effusion is reported to improve the survival rates.

In this case we reported a fetus who has hydrops fetalis(bilateral primary severe hydrotorax, ascite and skin edema) at 16 weeks and 4 days of gestation. Genetic analysis revealed mutations in the PIEZO1 and NF1A genes. It's shown that PIEZO1 gene is related with edema and perinatal fetal hydrops. Due to progressive fluid accumulation, bilateral thoracoamniotic shunts were placed. Follow-up imaging confirmed shunt functionality and resolution of hydrothorax. At term, a 3460-gram female infant with APGAR scores of 8 and 9 was delivered by ceaserian section. Postnatally, the shunts were removed, and the neonate remained clinically stable. The infant was discharged in good health with no complications.

Keywords: hydrops fetalis, PIEZO1 gene mutation,thoracoamniotic shunt

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29 Ekim - 1 Kasım 2025
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SS-062

Dandy-Walker malformation prenatal diagnosis and perinatal outcomes: three-year single-center case series

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Dandy-Walker malformation; prenatal diagnosis; perinatal outcomes

OBJECTIVE: Dandy-Walker malformation (DWM) is a rare congenital anomaly within the spectrum of cystic posterior fossa malformations. This study aimed to evaluate prenatal diagnosis and perinatal outcomes based on a three-year single-center experience.

MATERIALS-**METHODS**: Four cases diagnosed prenatally with DWM between 2022 and 2025 were retrospectively reviewed.

RESULTS: The mean maternal age was 29.5 years; mean gestational age at diagnosis was 26.8 weeks; mean gestational age at delivery was 37.0 weeks; and mean birth weight was 3121 g. Median Apgar scores were 6 at 1 minute and 9 at 5 minutes. One case was a dichorionic diamniotic twin pregnancy conceived via in vitro fertilization. Associated anomalies included ventriculomegaly, lobar holoprosencephaly, fetal growth restriction, and pelviectasis. Fetal MRI supported the diagnosis in two cases. All pregnancies were delivered by cesarean section.

CONCLUSION: Prenatal diagnosis of DWM is critical for pregnancy management and counseling. Our series emphasizes the diagnostic contribution of ultrasonography and fetal MRI, and the prognostic importance of associated anomalies.

Keywords: Dandy-Walker malformation, perinatal outcomes, prenatal diagnosis

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SS-063

İniensefali Nadir Vaka Sunumu

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[1]Etlik Şehir Hastanesi, Perinatoloji,Ankara
[2]Nurdağı Devlet Hastanesi,Kadın Hastalıkları ve Doğum,Gaziantep

GİRİŞ: İniensefali, nadir görülen bir gelişimsel anomalidir. Bu anomali, embriyolojik gelişim sırasında embriyonik servikal retrofleksiyonun devam etmesi ve servikal omurga veya üst toraks bölgesinde nöral oluğun kapanmaması sonucu gelişimin durmasından kaynaklanır.

Nadir görülmesi ve birinci ve ikinci trimesterin başlarında tespit edilebilmesi nedeniyle, iniensefali olan fetüsler nadiren canlı doğar.

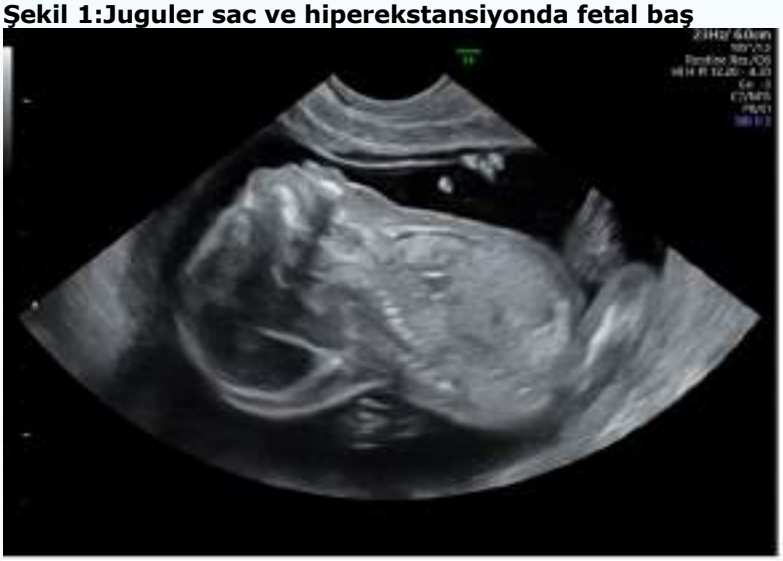
AMAÇ: Birinci ve ikinci trimester ultrasonografisinin erken tanı ve yönetimdeki rolünün vurgulanması ve ilk trimester rutin kontrolleri sırasında ultrasonografik tanı alan vakanınsunumu amaçlanmıştır. BULGULAR: Sonografik tanı,12-13.haftalarda konulabilir.Foramen magnum'u içeren oksiputta bir defekt, tüm vertebranın retrofleksiyonu, bu da fetüsün oksiputunu lomber bölgeye doğru yönlendirerek yukarı bakmasını zorlar. Vakaların yüzde 50'sinde değişen derecelerde açık omurga kusurları mevcuttur. İlişkili anomaliler vakaların yüzde 84'ünde görülür. Bunlar hidrosefali, mikrosefali, ventriküler atrezi, holoprosensefali, polimikroji, serebellar vermis agenezi, oksipital ensefalosel, diyafragma hernisi, toraks deformiteleri, üriner sistem anomalileri, yarık dudak ve damak, omfalosel ve polihidromnios gibi bulgular olabilir.

41 yaş G1 olan hasta 14. Gebelik haftasında rutin kontrol için polikliniğe başvurdu.Özgeçmişinde geçirilmiş operasyon ve bilinen ek hastalık yoktu.Hastanın yapılan ultrasonografisinde duktus venozusta ters a dalgası izlendi.Böbreklerde hiperekojenite, cilt altı ve scalp ödemi izlendi.Bilateral juguler sac ve iniensefali izlendi.Genetik tanı testi hakkında bilgilendirilen aile test istemedi.Terminasyon seçeneği sunuldu ve kabul etti. Medikal abortus ile gebelik sonlandırıldı.

SONUÇ: Erken hafta ultrasonografik tarama dikkat ile yapılmalıdır.Erken haftalarda ultrasonografinin tanı gücü gün geçtikçe artmaktadır.İniensefali nadir görülen lethal bir yapısal anomalidir.Erken hafta taramalarda akılda tutulması ayırıcı tanı için önemlidir.

Anahtar Kelimeler: Erken hafta ultrasonografi, İniensefali, Terminasyon

Şekil 1:Juguler sac ve hiperekstansiyonda fetal baş



Şekil 1:Juguler sac ve hiperekstansiyonda fetal baş

Şekil 2: Abort materyali

Şekil 2: Abort materyali

Şekil 3: Abort materyali

Şekil 3: Abort materyali

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SS-064

Sonographic Appearance And Intraoperative Confirmation Of A Silent Uterine Scar Dehiscence

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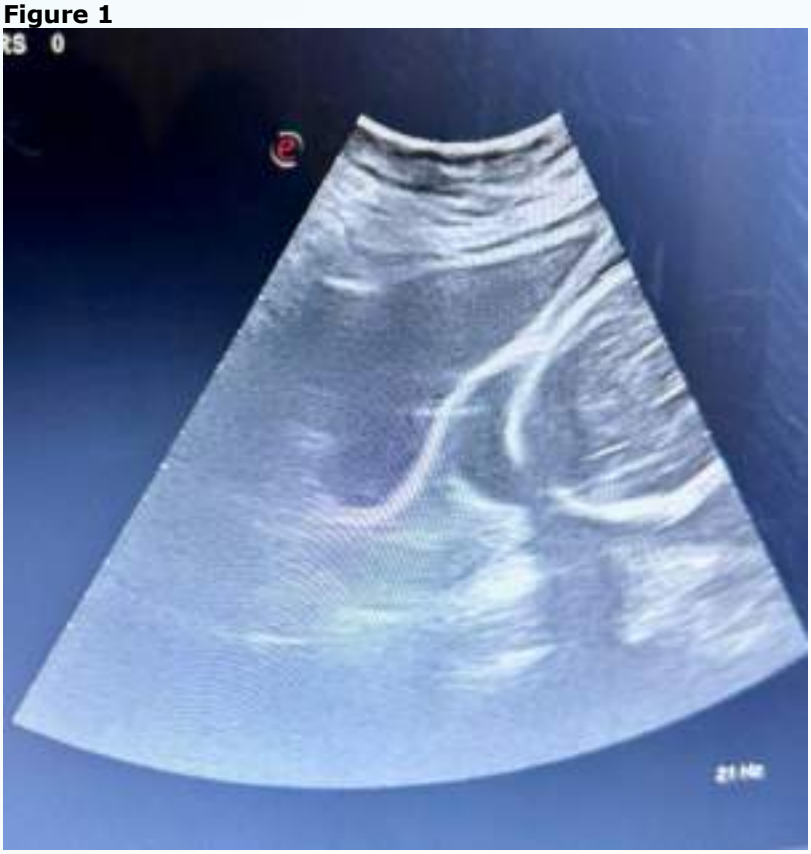
[2]Istanbul Training and Research Hospital, Istanbul, Turkey

Uterine scar dehiscence is partial separation of uterine myometrium following a previous cesarean section, with the serosal layer remaining intact. It is usually asymptomatic and has a reported incidence ranging from 0.2% to 4.6% in retrospective studies. Scar defects are detected using ultrasonography in 4–7% of pregnancies during the later weeks of gestation. If the scar defect is not diagnosed, it may progress to complete uterine rupture, leading to significant maternal-fetal morbidity and mortality. We present a case of a 28-year-old gravida 2, para 1 patient with a history of cesarean section. Termination of pregnancy was planned due to multiple fetal anomalies, including agenesis of corpus callosum with coexisting findings such as colpocephaly, absence of cavum septum pellucidum and cardiac muscular ventricular septal defect, in conformity with parental will. Misoprostol protocol was initiated in accordance with FIGO 2023 guidelines. Although effective uterine contractions were induced, due to poor advancement in cervical Bishop's score, a follow-up ultrasonographic evaluation was performed, revealing a discontinuity in the muscular layer of the anterior uterine wall. Cesarean section was promptly carried out, and intraoperative findings confirmed the presence of uterine scar dehiscence. This case highlights the importance of ultrasonographic evaluation of the uterine scar in post-cesarean pregnancies, particularly when labor fails to progress. Monitoring the integrity of the uterine myometrium can provide critical information that may alter clinical management and prevent severe complications. Routine consideration of scar site assessment may be suggested during the process of medical termination in patients with prior uterine surgery.

Keywords: Cesarean section, ultrasonography, uterine rupture,

Figure 1

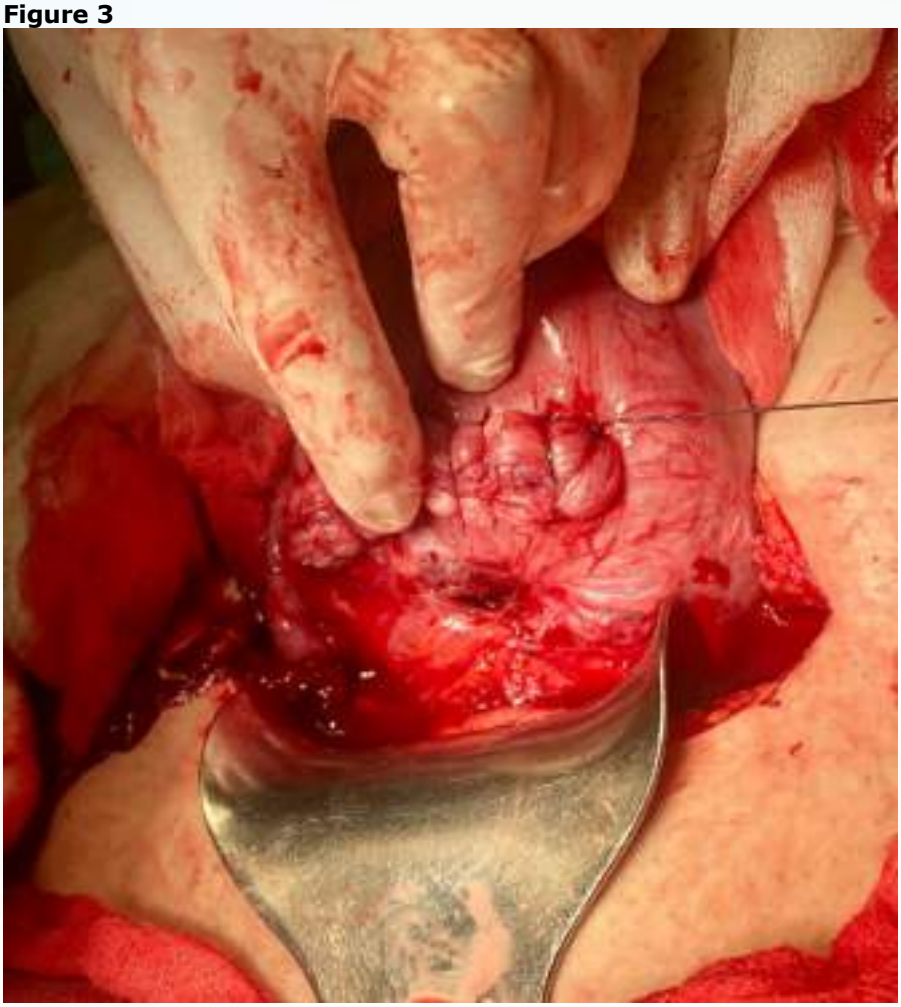


Transabdominal Detection of Uterine Dehiscence

Figure 2

Transvaginal Detection of Uterine Dehiscence

Figure 3



Intraoperative Detection of Uterine Dehiscence

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SS-065

Amniotic Band Syndrome: A Case Series Highlighting Prenatal Diagnostic Challenges

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INTRODUCTION: Amniotic band syndrome (ABS) is a rare congenital anomaly with a broad spectrum of clinical manifestations, ranging from mild constriction rings to severe craniofacial and limb defects. Early prenatal detection is critical for prognosis, parental counseling, and pregnancy management. We present a case series of three fetuses with ABS diagnosed prenatally, emphasizing the variability of clinical presentation and the importance of multidisciplinary evaluation.

Cases
Case 1: A 36-year-old woman, primigravida following in vitro fertilization, was diagnosed at 11 weeks with absence of the left humerus, radius, ulna, and hand, along with an amniotic band extending from the cervix. Additional findings included hyperechogenic bowel and subcutaneous edema. The pregnancy was terminated at 14 weeks.

Case 2: A 28-year-old primigravida at 15 weeks showed asymmetry of cerebral hemispheres and a midline facial defect on ultrasound. Findings were attributed to ABS. The pregnancy was terminated at parental request; genetic analysis revealed no abnormalities.

Case 3: A 27-year-old multigravida with prior abortions was found at 29 weeks to have fetal edema of the left hand and absence of the right middle finger distal phalanx. An amniotic band encircling the cord was observed. Elective cesarean delivery at 34 weeks resulted in a live female neonate with limb deformities.

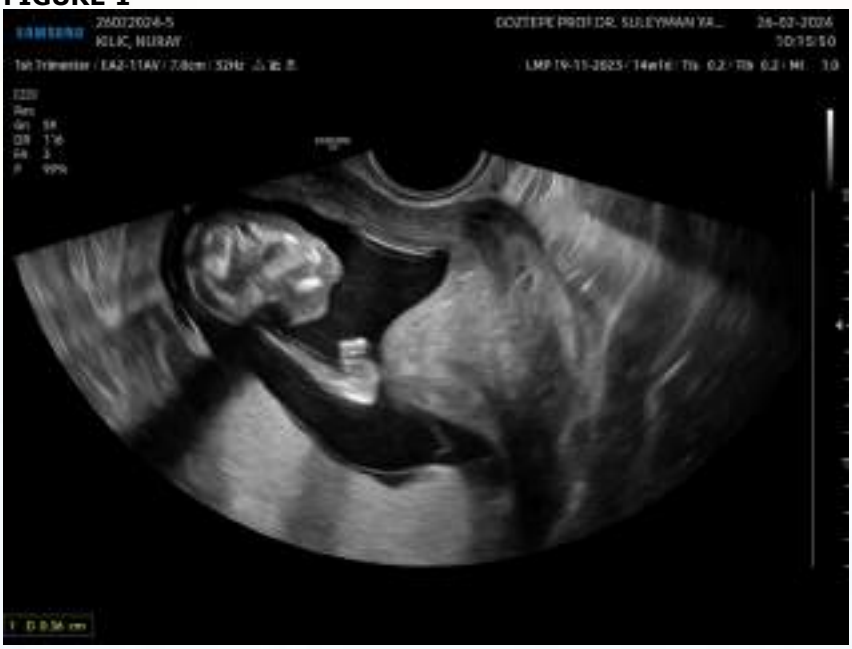
CONCLUSION: This case series illustrates the diverse spectrum of ABS and its diagnostic challenges. Prenatal ultrasonography plays a crucial role in early recognition, prognosis, and counseling. Multidisciplinary management is essential to optimize perinatal outcomes and provide appropriate family-centered care.

Keywords: Amniotic band syndrome, prenatal diagnosis, congenital anomalies, ultrasonography, perinatal outcome

Fıgıre 4

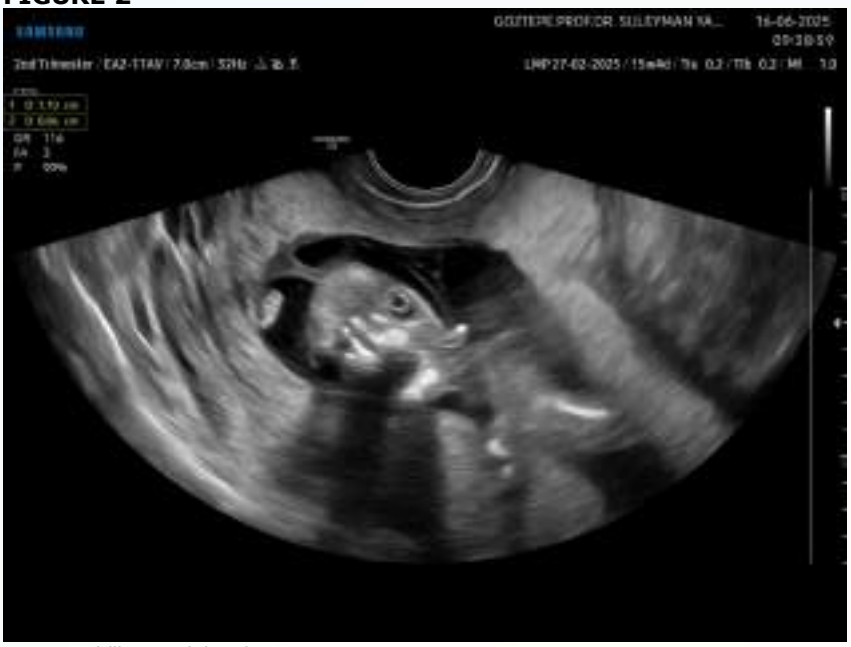
Case 3 Amniotic band encircling the umbilical cord.

FİGURE 1



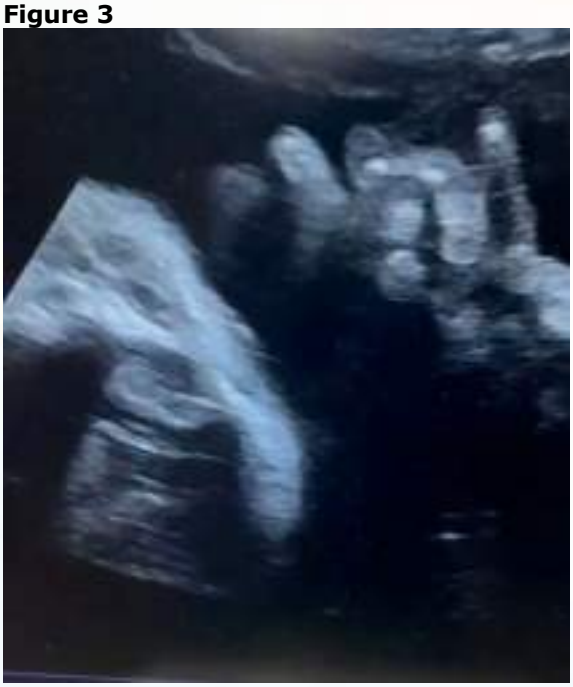
Sagittal obstetric ultrasound image of a 14 weeks +1 day fetus. The right upper extremity is not visualized distal to the elbow, and the hand appears to be deviated. The left arm is not clearly visible in this image, limiting evaluation. The findings are consistent with right radial aplasia.

FİGURE 2



Case 2 Midline Facial Defect

Figure 3



Case 3 Fetal edema of the left hand and absence of the distal phalanx of the right middle finger.

SS-066

Giant Lymphangioma of a Fetus: A Case Report

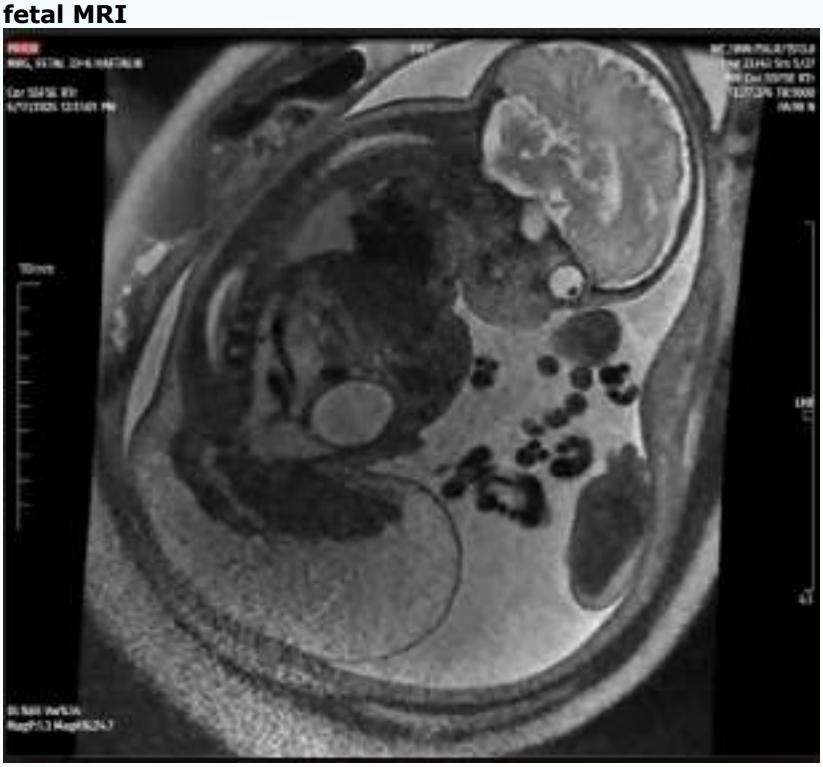
Ozlem Aldemir Bukagikiran, Mucize Eric Ozdemir
ZEYNEP KAMİL KADIN VE ÇOCUK HASTALIKLARI EĞİTİM VE ARAŞTIRMA HASTANESİ

Lymphangioma (lymphatic malformation), is a vascular malformation that typically develops during the second and third trimesters of gestation and often demonstrates progressive and infiltrative growth. The incidence is estimated to range from 0.8 to 18 per 10,000 fetuses. Lymphangiomas vary in size, from a few millimeters to more than 10 cm in diameter, and may present as unilocular or multilocular lesions. Prenatally, lymphangiomas are usually asymptomatic; however, they can exert a mass effect on adjacent structures depending on their location.

We report the case of a 26-year-old primigravida who was referred at 28 weeks of gestation with a preliminary diagnosis of sacrococcygeal teratoma. Ultrasound revealed polyhydramnios and a septated anechoic cystic mass extending along the left lateral abdominal wall, from the renal level to the sacral region, compressing the left kidney and stomach. The lesion also extended from the gluteal region along the left femur to the knee. Fetal MRI demonstrated a multilocular, predominantly cystic lesion in the left hemiabdomen. The mass extended from the presacral area through the posterior aspect of the right thigh to the proximal crus, infiltrating between muscle planes, and reaching the subcutaneous tissue at the lumbosacral level. These findings were consistent with a veno-lymphatic malformation. The baby was delivered via C-section at term, weighing 3880 grams. Postnatal MRI findings were consistent with the prenatal imaging results. Definitive postnatal management consisted of sclerotherapy using bleomycin in combination with oral sirolimus. Genetic testing, including cytogenetic and chromosomal microarray analysis, was performed and revealed normal results.

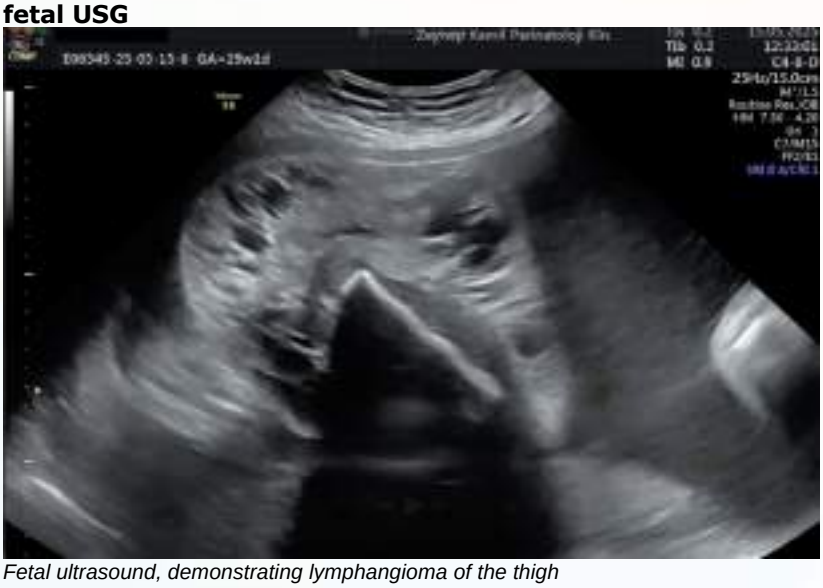
Keywords: fetal lymphangioma, lymphatic malformation, cystic mass

fetal MRI



Fetal MRI, demonstrating the lymphangioma-from gluteal region along the fetal thigh.

fetal USG



Fetal ultrasound, demonstrating lymphangioma of the thigh

newborn

The newborn’s left thigh appeared enlarged due to the lymphangioma.

SS-067

Arthrogryposis Multiplex Congenita: A Rare Case from Prenatal Diagnosis to Genetic Confirmation

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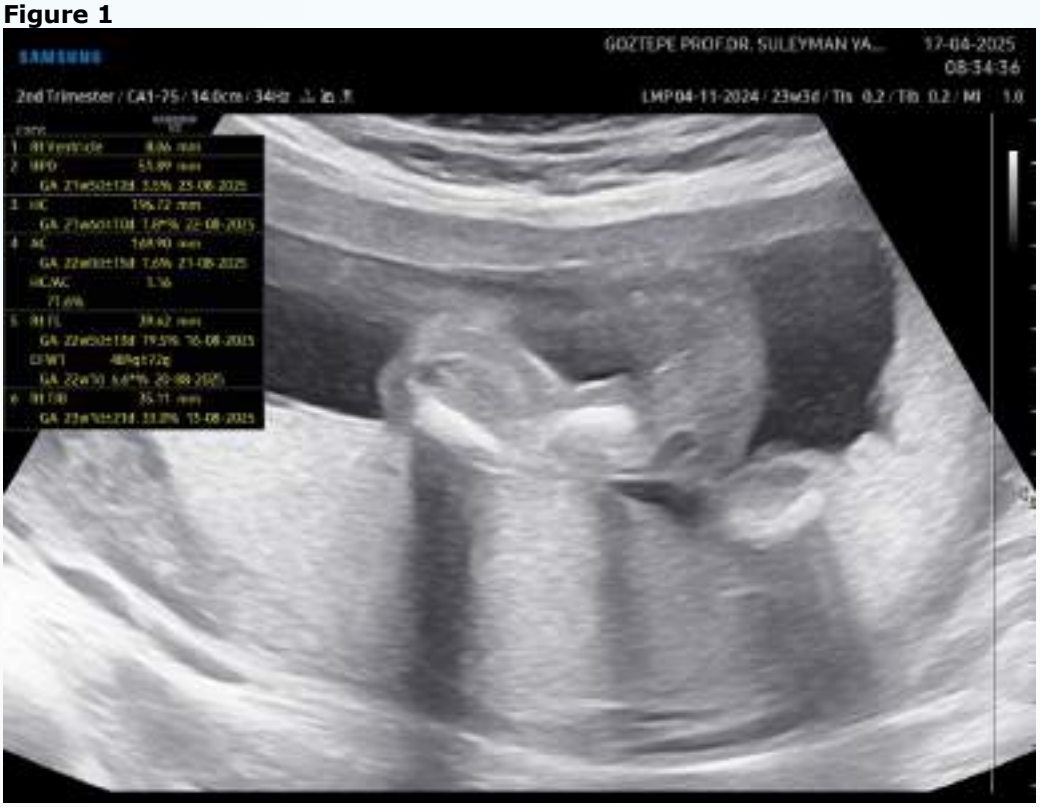
OBJECTIVE: Arthrogryposis multiplex congenita(AMC) is characterized by joint contractures involving at least two different body regions of the fetus. It is a heterogeneous syndrome group with an estimated prevalence of approximately 1 in 3000 live births, which is higher than many other conditions routinely screened in the prenatal period. In this report, we present a case of AMC diagnosed during the prenatal period, in which genetic mutations were confirmed through cordocentesis.

CASE: 27y, G4P1A2, 23w pregnant patient was referred for second-trimester anomaly screening. The parents were consanguineous, and no prior screening tests had been performed. Ultrasonography revealed flexion deformities of the hands and feet, bilateral pes equinovarus, microcephaly, nasal bone hypoplasia, and micrognathia (Figure 1-3). The cavum septi pellucidi was absent, the optic chiasm measured below the 3rd percentile, and the anterior horns of the lateral ventricles were fused at the midline. Parieto-occipital sulcus and insular development were delayed for gestational age. Cordocentesis was performed, and CES revealed VUS in the NALCN and MYH3 genes associated with AMC. Following detailed genetic counseling regarding prognosis and quality of life, the family opted for termination. Postnatal evaluation confirmed multiple contractures (Figure 4).

CONCLUSION: The prognosis of AMC varies according to etiology, severity of contractures, and associated anomalies.Early and accurate diagnosis improves detection rates and supports timely counseling for families.

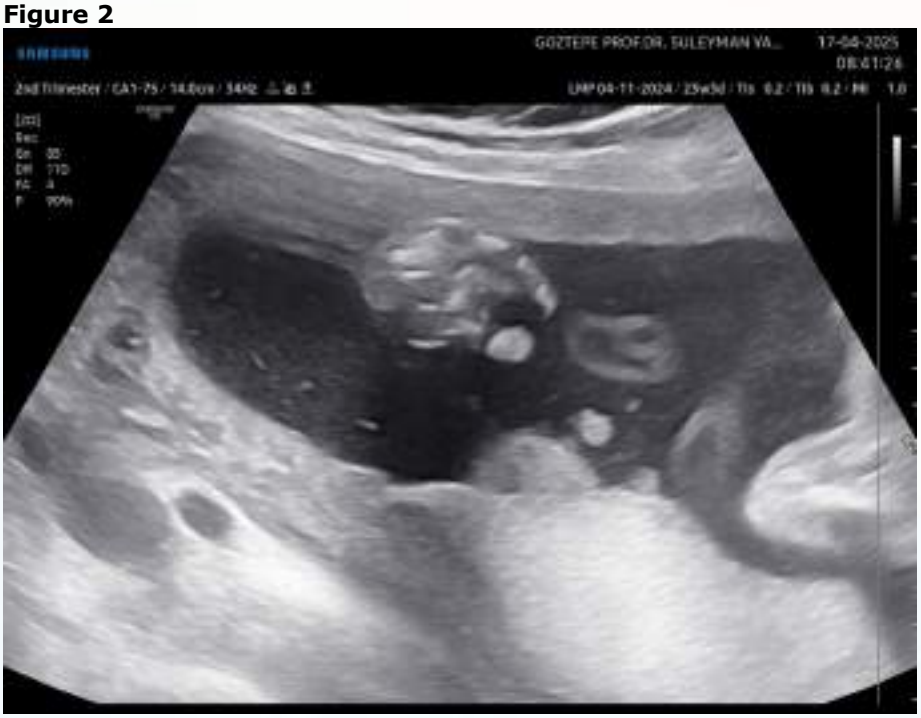
Keywords: Arthrogryposis multiplex congenita, Arthrogryposis, Cordocentesis, Prenatal diagnosis

Figure 1



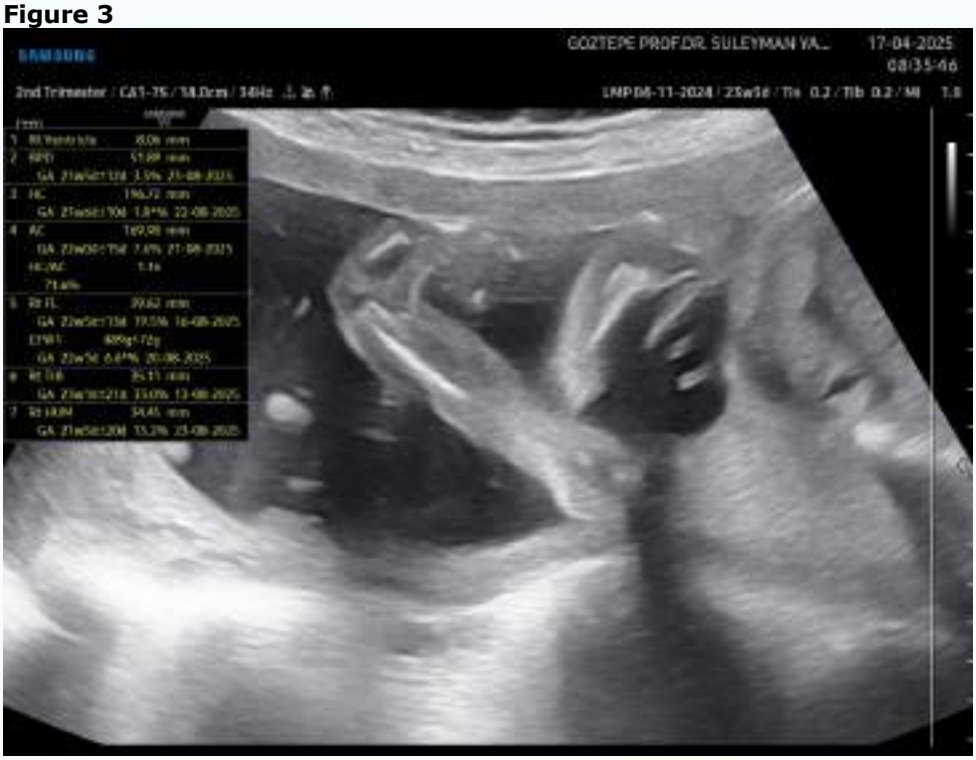
Pes equinovarus

Figure 2



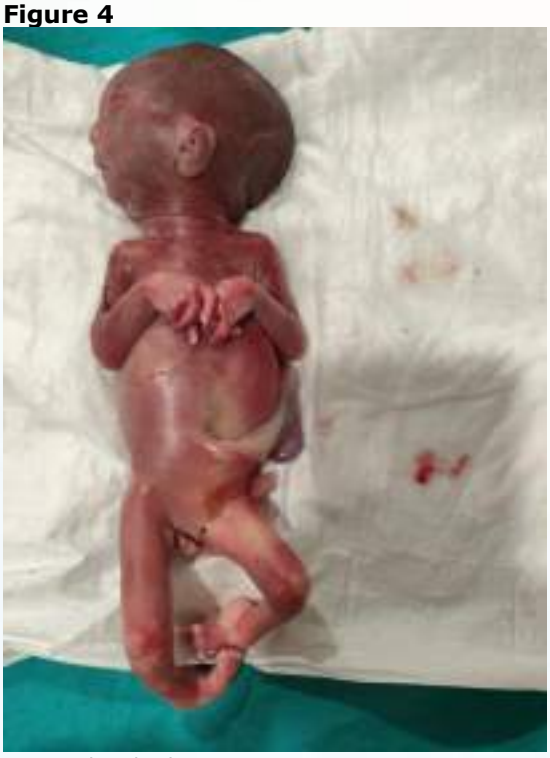
Flexion deformities of the hands

Figure 3



Flexion contractures in the hands and wrists

Figure 4



Postnatal evaluation

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SS-068

Prenatally Detected Fetal Adrenal Hematoma: A Case Report

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[2]Department of Perinatology, Istanbul Medeniyet University, Istanbul, Turkey

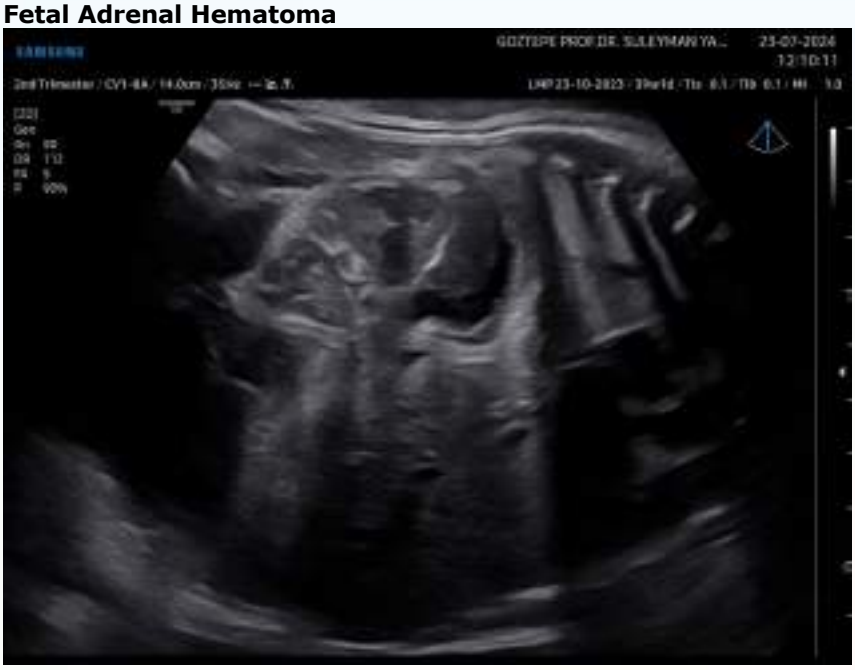
INTRODUCTION: Fetal adrenal hematoma is a condition rarely diagnosed in the antenatal period. With routine prenatal ultrasonography, detection of fetal adrenal masses has increased, but determining the origin of echogenic lesions remains crucial. Differential diagnosis includes neuroblastoma, extralobar pulmonary sequestration, lymphatic malformation, duplex kidney, and adrenal hyperplasia. Serial ultrasonography and postnatal multidisciplinary evaluation are critical. We present the favorable outcome of a prenatally detected adrenal hematoma managed conservatively.

CASE: A 27-year-old gravida 2, para 1 woman with type 2 diabetes at 39+1 weeks' gestation was found to have polyhydramnios and an incidental left fetal adrenal hematoma measuring 30x15 mm on ultrasonography. After prenatal counseling, a male infant weighing 3290 g with Apgar scores of 7 and 9 was delivered by cesarean section. Postnatal ultrasonography revealed a 20×10 mm cystic lesion in the left adrenal region containing a coagulum. Laboratory findings were normal, with no signs of adrenal insufficiency. Serial ultrasonography showed progressive regression, with complete resolution by the 5th month. During follow-up, multiple renal stones appeared in the left kidney, without metabolic abnormalities, which resolved by the 9th month. At 14 months, the infant demonstrated normal development without additional pathology.

CONCLUSION: Prenatally detected adrenal hematomas should be differentiated from malignant masses, and parents should be counseled regarding antenatal findings and delivery planning. Most lesions regress spontaneously and can be managed through ultrasonographic monitoring and multidisciplinary care. This case highlights that adrenal hematomas do not necessarily require early postnatal surgical or interventional procedures and usually show favorable prognosis under appropriate surveillance.

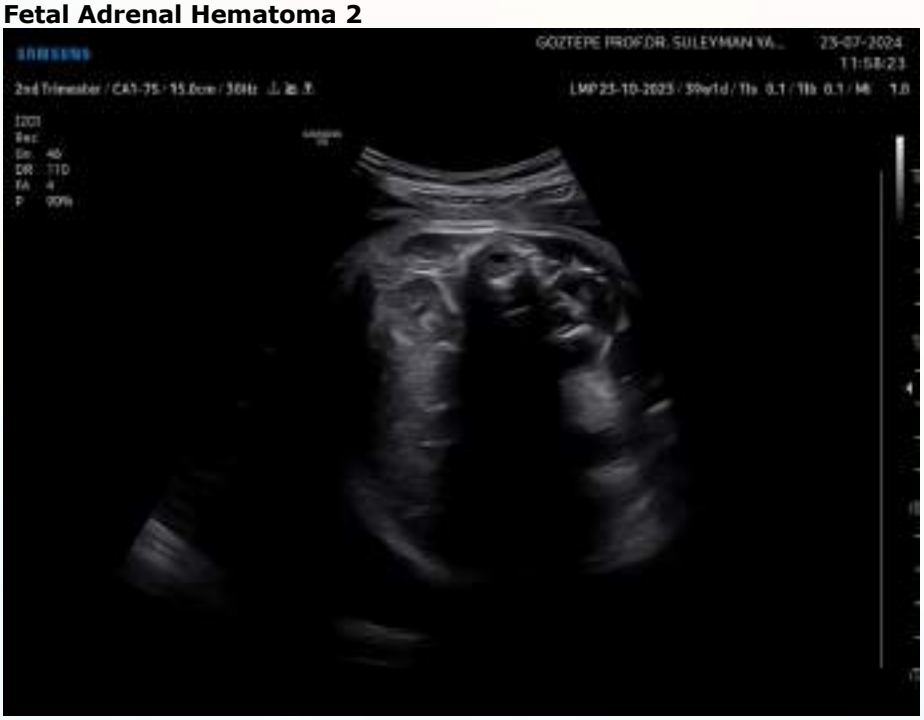
Keywords: fetal adrenal hematoma, prenatal diagnosis, ultrasonography

Fetal Adrenal Hematoma



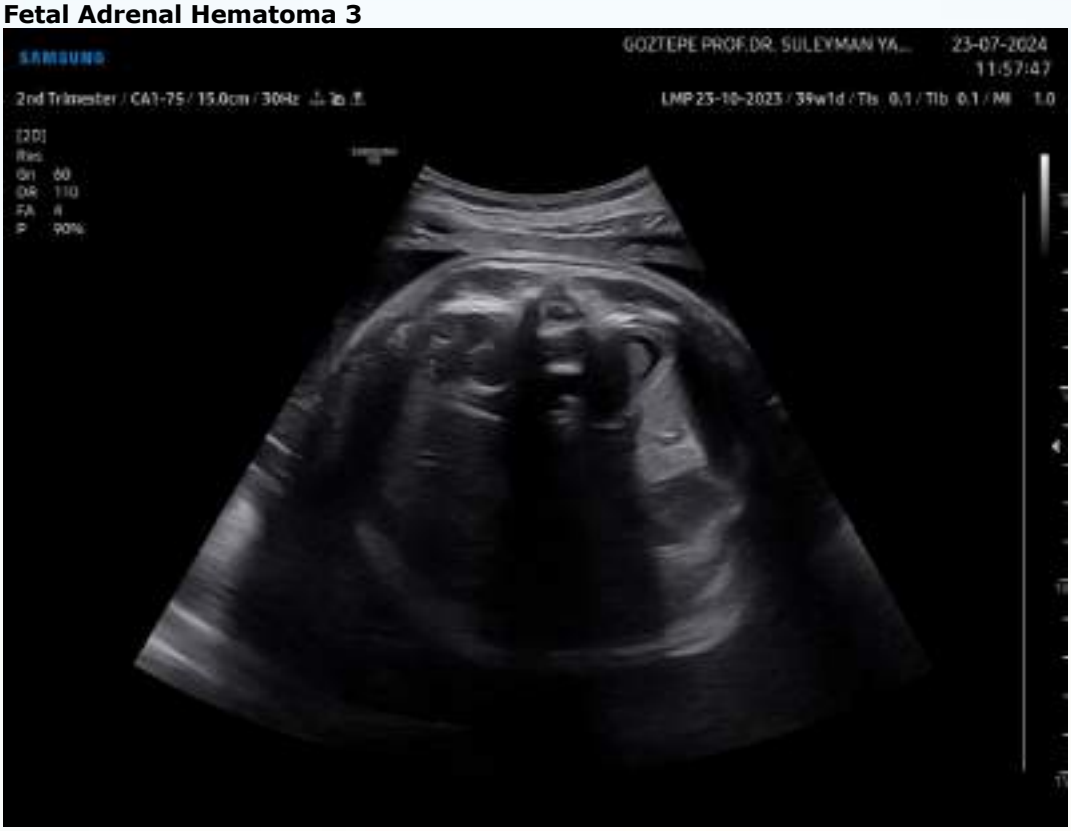
Sagittal US plane of fetal adrenal hematoma

Fetal Adrenal Hematoma 2



Axial US plane of fetal adrenal hematoma

Fetal Adrenal Hematoma 3



Axial US plane of fetal adrenal hematoma

SS-069

Prenatal Detection of Binder Syndrome-Like Facial Profile: A Case Report

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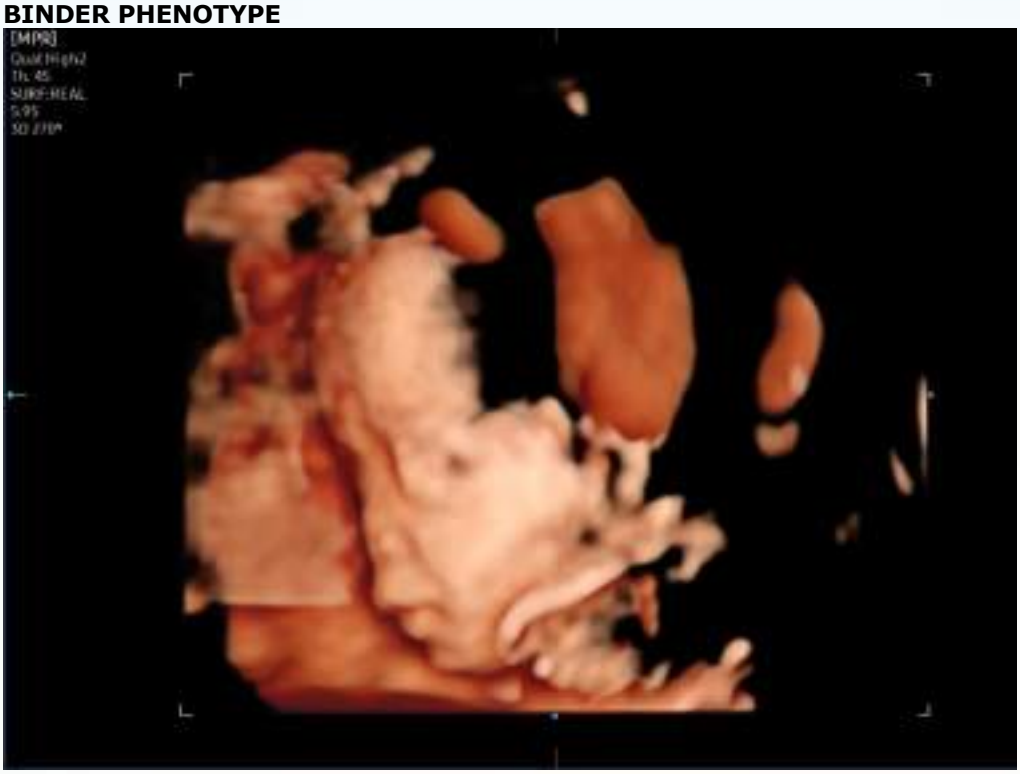
INTRODUCTION: Binder syndrome, also referred to as nasomaxillary hypoplasia or maxillonasal dysplasia, is a rare congenital craniofacial anomaly characterized by underdevelopment of the anterior maxilla and nasal structures. This condition results in distinctive craniofacial features, including a concave or retruded midface, a broad and flattened nasal bridge, a shortened columella, a prominent upper lip, and frequently Class III dental malocclusions. These characteristic findings often allow for early and accurate diagnosis of the syndrome during prenatal or postnatal evaluations.

Case Presentation: A 36-year-old G4P2CS1 pregnant woman presented at 23+2 weeks of gestation, when a second-trimester anomaly scan revealed a flattened fetal facial profile on ultrasonography. No other significant structural abnormalities were detected. The first-trimester combined screening had shown a low-risk result. Given the suspicion of a Binder-like phenotype, amniocentesis was performed. FISH analysis demonstrated suspicious signals suggestive of trisomy 18(1%).Subsequent conventional karyotyping revealed a normal chromosomal complement of 46, however, trisomy of chromosome 18 was observed in 1 of 100 metaphases, which was interpreted as consistent with level 1 mosaicism. WES did not reveal any pathogenic variants. Since level 1 mosaicism is often associated with pseudomosaicism and isn't expected to be reflected in the fetus, advanced cordocentesis wasn't performed. The postpartum evaluation revealed no additional abnormalities.

CONCLUSION: Suspicious facial profile findings should be further evaluated with advanced imaging techniques and comprehensive genetic testing. A multidisciplinary approach is essential for establishing an accurate diagnosis and guiding prenatal counseling, pregnancy management, and informed decision-making.

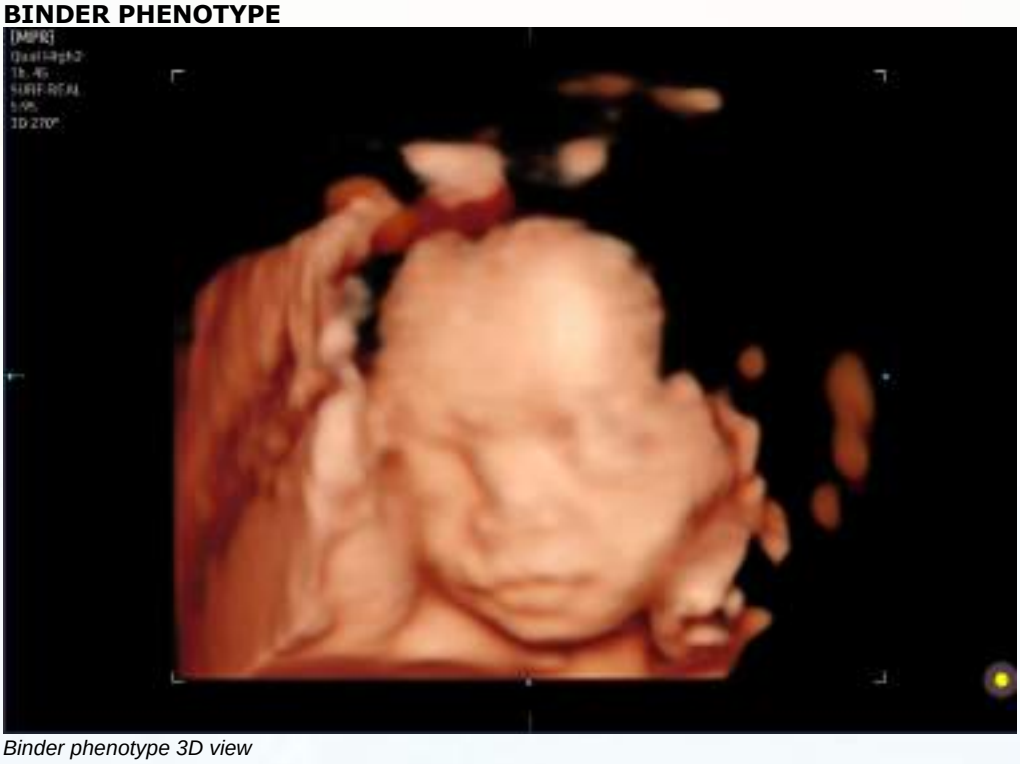
Keywords: Binder syndrome, Facial profile, Maxilla

BINDER PHENOTYPE



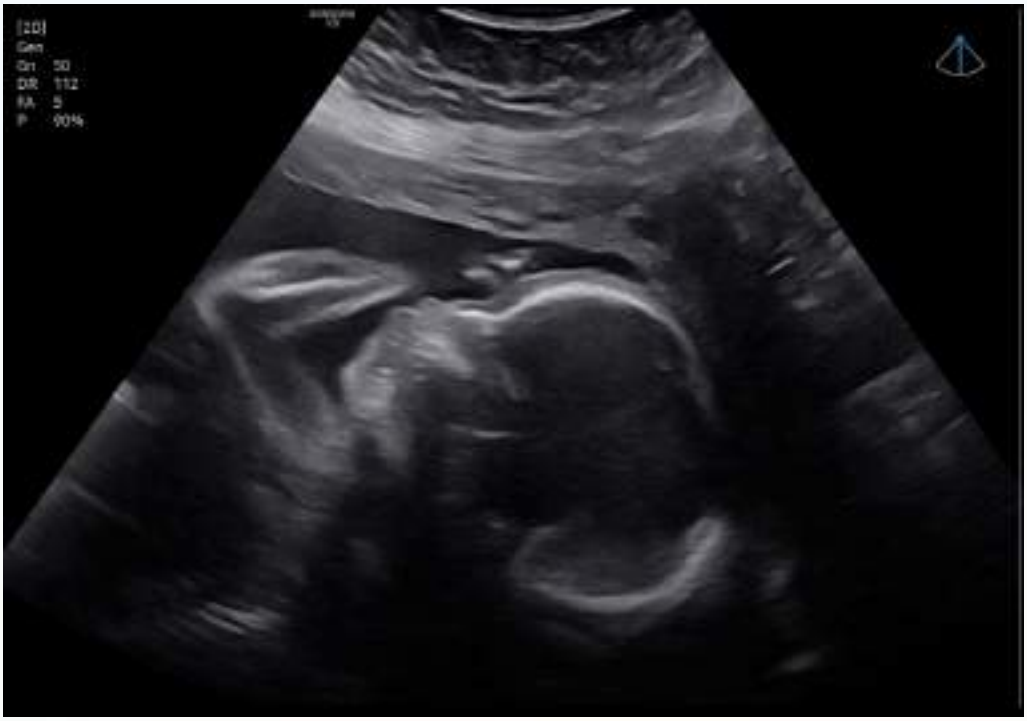
Binder Phenotype 3d view

BINDER PHENOTYPE



Binder phenotype 3D view

BINDER PHENOTYPE



Binder Phenotype 3D view

SS-070

Pregnancy in a Patient with Takayasu Arteritis Complicated by Pulmonary Hemorrhage: A Case Report

Esra Nuhoğlu Bilgin, Pınar Tokdemir Çalış, Hatice Aydın Salman, Arzu Sümer Ortaç, Esin Şahin Toruk, Bilal Emre Erzeneoğlu, Deniz Karçaaltıncaba, Tuncay Nas
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Background Takayasu arteritis (TA) is a rare, chronic granulomatous vasculitis affecting large vessels, primarily the aorta and its major branches. It predominantly affects women of reproductive age. Pregnancy in patients with TA is associated with increased maternal and fetal risks, including hypertension, vascular stenosis, thrombosis, and placental insufficiency. Although pulmonary involvement is reported in approximately 40% of TA patients, pulmonary hemorrhage and hemoptysis remain extremely rare yet life-threatening complications.

A 33-year-old G3A1P1 woman with a 15-year history of TA presented hemoptysis following a coughing episode at 21 weeks of gestation. She had prior aorto-biiliac and thoracoabdominal bypass surgeries. Certolizumab, a TNF-alpha inhibitor, had been self-discontinued 3 months earlier. Thoracic CT angiography ruled out pulmonary embolism. After stabilization of bleeding bronchoscopy was performed.

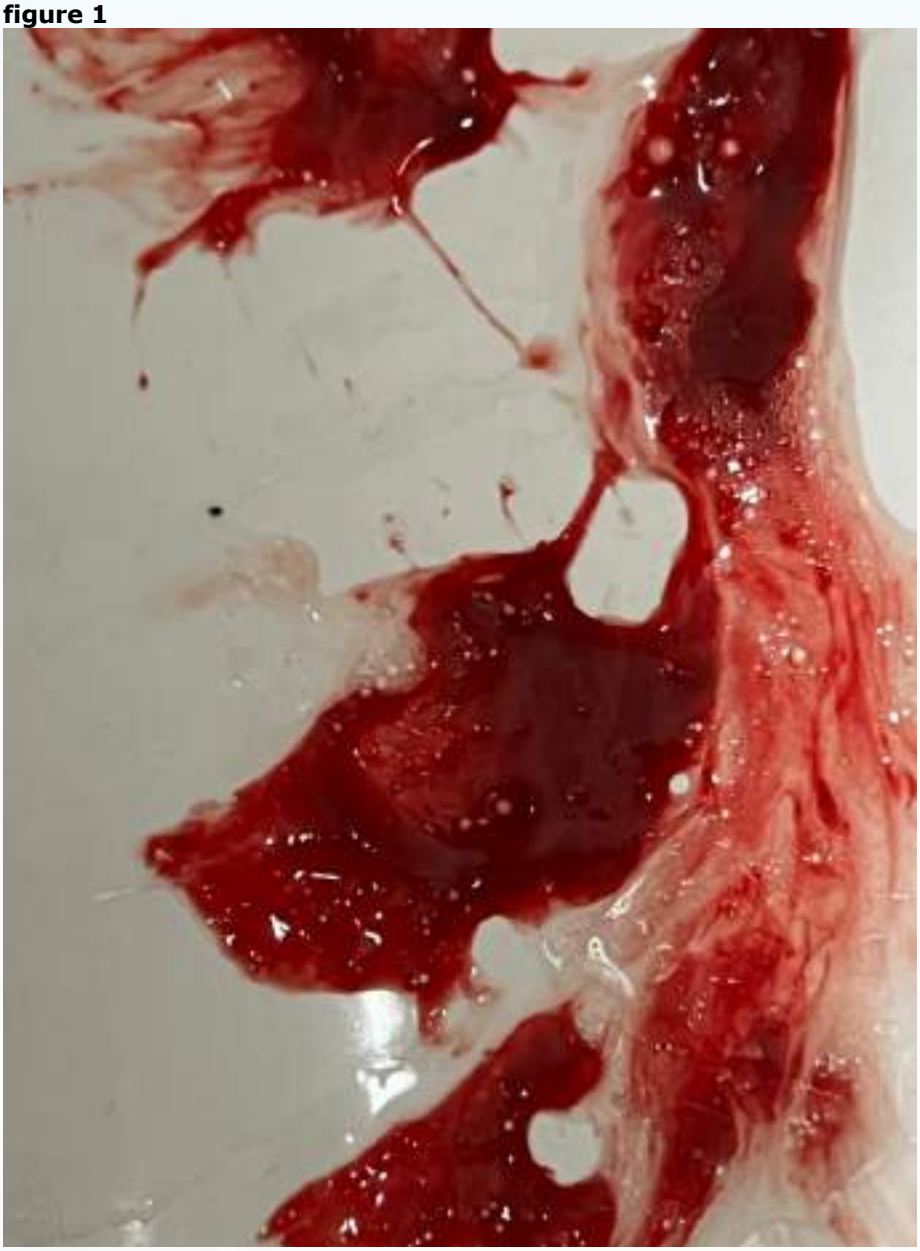
Given stability and absence of new vascular findings, conservative treatment was pursued. No recurrence of hemoptysis occurred. She was delivered a healthy baby at 37+1 weeks by planned cesarean section since she had previous cesarean section.

This case underscores the complexity of managing TA in pregnancy, particularly when complicated by rare events such as pulmonary hemorrhage. Physiological hemodynamic changes during pregnancy—including increased blood volume and cardiac output—may exacerbate underlying vascular fragility in patients with TA, increasing the risk for vascular complications. In the presented case, a history of extensive vascular surgery, prior thrombophlebitis, and recent discontinuation of biologic therapy represented multiple risk factors for disease reactivation or vascular instability.

This case emphasizes the importance of individualized and multidisciplinary management in pregnant patients with TA.

Keywords: Takayasu Arteritis, pregnancy, pulmonary hemorrhage

figure 1



Hemoptysis of the case

SS-071

Silent placental abruption leading to severe fetal anemia and hydrops resulting in sudden fetal deterioration

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Background
Placental abruption complicates approximately 0.4–1.0% of pregnancies typically presenting with vaginal bleeding, and abdominal-uterine tenderness. Although cardiotocography (CTG) may remain reactive in mild cases, with progression, recurrent late decelerations, reduced variability, sustained bradycardia, sinusoidal pattern, or even fetal asystole may be encountered. However, up to 10–20% of cases may remain clinically silent, being recognized only postpartum or after unexplained fetal distress.

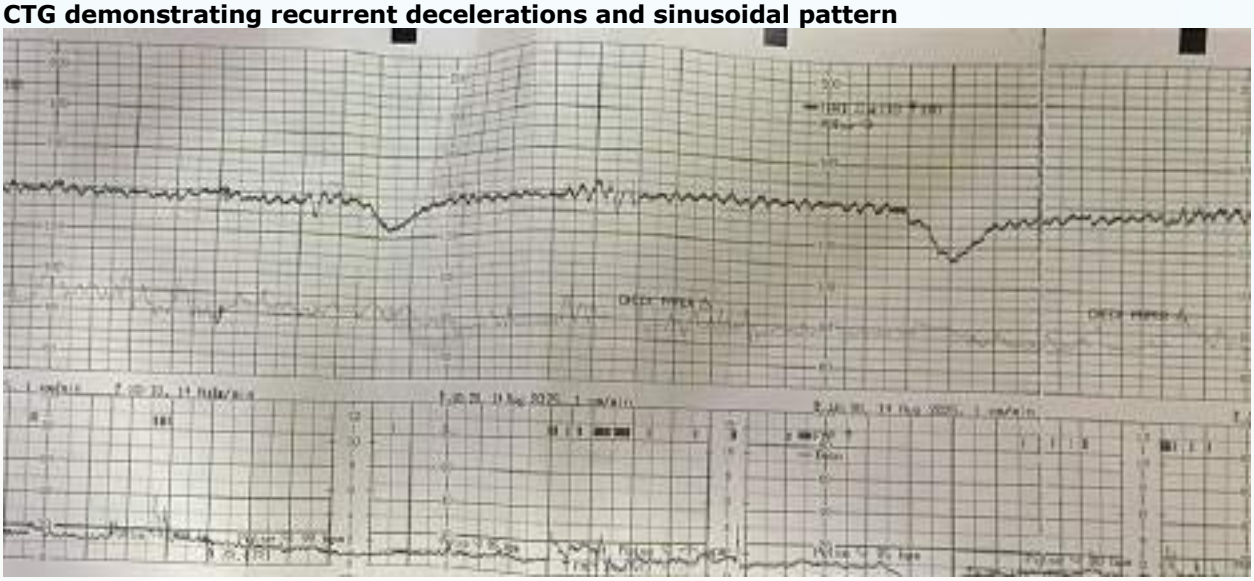
Case Report A 25-year-old gravida 4, para 2 patient at 38th week, otherwise asymptomatic, presented with decreased fetal movements. Ultrasound exam revealed fetal ascites and subcutaneous edema accompanied by oligohydramniosis and subchorionic hematoma. CTG demonstrated recurrent decelerations and sinusoidal pattern. Emergency cesarean delivery was performed due to non-reassuring findings. A live female infant weighing 2880 g with Apgar scores of 2-5 was delivered. The newborn was found to be severely anemic with Hb: 2,9 gr/dL.

Management and Outcome Maternal and newborn assessment for the etiology of fetal anemia were unremarkable. Placental pathology confirmed infarction, hemorrhage, and congestion. The mother recovered uneventfully and was discharged on postoperative day 2. The neonate at birth presented with pallor, tachypnea, and absence of spontaneous respiration, requiring intubation. She underwent daily transfusions and achieved stabilization with hemoglobin 13.1 g/dL at post-partum 7th day.

CONCLUSION: Silent placental abruption may sometimes lead to severe fetal anemia and related consequences. Sudden deterioration of a fetus with specific clinical (decreased movements), ultrasound (hydrops) and CTG (sinusoidal pattern) findings in an otherwise asymptomatic mother should alert the clinician about the rare cases of silent ablation.

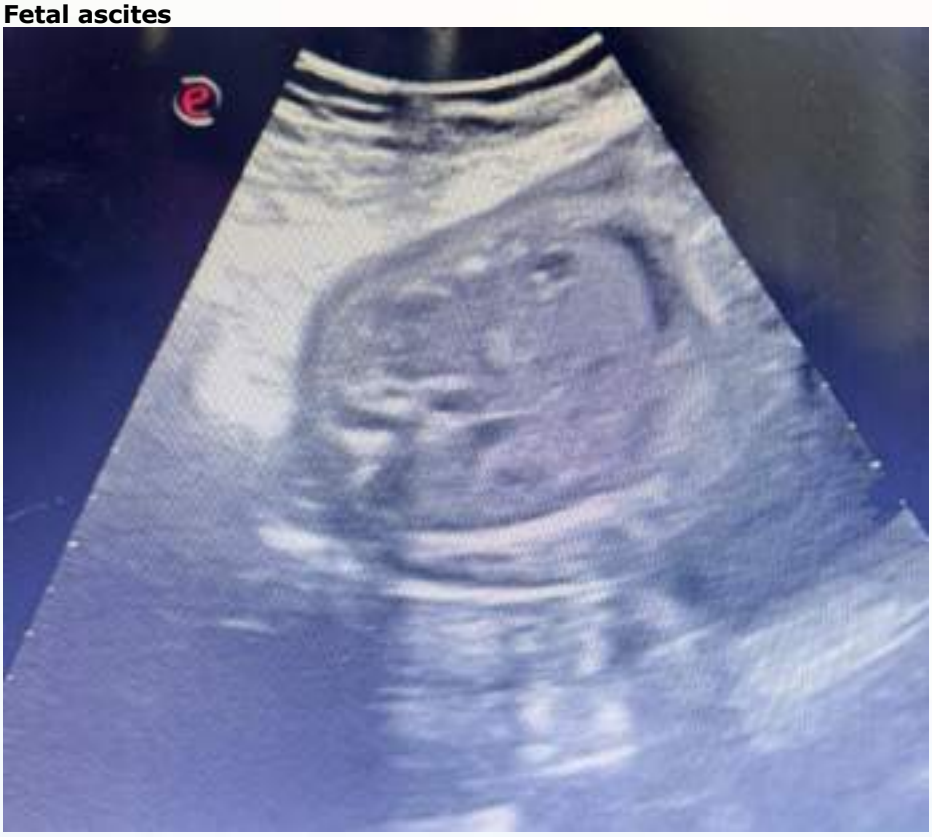
Keywords: Fetal anemia, silent placental abruption, sinusoidal CTG

CTG demonstrating recurrent decelerations and sinusoidal pattern



CTG demonstrating recurrent decelerations and sinusoidal pattern

Fetal ascites



fetal ascites

SS-072

"Prenatal Diagnosis of Prepontine Arachnoid Cysts: Report of Two Cases and Review of the Literature

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Department of Perinatology, Etlik City Hospital, Ankara,Turkey

Objective We aimed to present two cases of prepontine arachnoid cyst diagnosed prenatally, supported by a review of the current literature. Case Presentation Case 1 was a 22-year-old gravida 2 woman. She had not undergone first- or second-trimester screening tests. She had no comorbidities or regular medication use. At 27 weeks of gestation, she was referred for evaluation. Ultrasound revealed a 21×9 mm cystic lesion in the posterior fossa. Fetal magnetic resonance imaging showed a cisterna magna anteroposterior diameter of 12 mm with asymmetric enlargement and mass effect on the cerebellar hemisphere. Findings were consistent with a prepontine arachnoid cyst. Case 2 was a 33-year-old gravida 2 woman. She was referred at 33 weeks of gestation. Ultrasound demonstrated an 11×10 mm cystic lesion consistent with a prepontine arachnoid cyst. No fetal MRI was performed in this case. Amniocentesis was recommended for both patients but declined. Prenatal counseling was provided, and both pregnancies are being followed with a multidisciplinary approach. Conclusion Arachnoid cysts are rare congenital anomalies of the central nervous system. Prenatal diagnosis is crucial for predicting prognosis, which depends on the size and location of the cyst. These two cases highlight the role of ultrasonography and MRI in prenatal diagnosis and emphasize the importance of multidisciplinary follow-up in the postnatal period.

Keywords: Prepontine arachnoid cyst, Fetal MRI, Cranial cyst

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29 Ekim - 1 Kasım 2025
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SS-073

A Rare Presentation of Neural Tube Defects: Prenatal Diagnosis of Isolated Frontal Encephalocele

Esin Şahin Toruk, Arzu Sümer Ortaç, Hatice Aydın Salman, Esra Nuhoğlu Bilgin, Bilal Emre Erzeneoğlu, Pınar Tokdemir Çalış, Deniz Karçaaltıncaba, Mehmet Zeki Taner
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INTRODUCTION: Encephalocele is a rare congenital neural tube defect characterized by the herniation of intracranial contents through a skull defect. Approximately 75% of cases occur in the occipital region, 15% in the frontal region, and the remainder in the parietal region. Its incidence ranges between 1:2000 and 1:5000 live births with no gender predilection. Frontal encephaloceles are rare and often involve craniofacial structures, with cosmetic and neurological implications. Prenatal ultrasonography is essential for diagnosis and counseling.

Case Presentation: A 40-year-old woman, gravida 4, para 2, with two living children, was referred at 14+5 weeks of gestation with a preliminary diagnosis of cystic hygroma. Her first pregnancy ended in termination for fetal hydrocephalus. There was no consanguinity or family history of genetic disorders, and the patient had not used folic acid during this pregnancy. Detailed ultrasonography revealed a midline frontal encephalocele associated with a skull defect. No vascular flow was observed within the lesion, and no additional anomalies were detected. The parents were counseled regarding the diagnosis, prognosis, and management. An invasive prenatal genetic test was performed, and FISH analysis revealed a normal result. Considering the findings and prognosis, termination was planned with parental agreement. Post-termination evaluation confirmed the prenatal diagnosis of frontal encephalocele.

CONCLUSION: Frontal encephalocele is a rare neural tube defect. Early diagnosis by prenatal ultrasonography is critical for prognosis and counseling. This case illustrates that frontal encephalocele may occur in isolation and highlights the diagnostic value of prenatal imaging. Reporting such rare cases increases clinical awareness.

Keywords: Frontal encephalocele, prenatal diagnosis, ultrasonography, neural tube defect

SS-074

Prenatal Management of Severe Familial Hypertriglyceridemia and Chronic Pancreatitis in Pregnancy Associated with APO A5 Homozygous Mutation: A Case Report

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OBJECTIVE: Hypertriglyceridemia-induced pancreatitis in pregnancy is a rare but potentially life-threatening condition for both mother and fetus. Genetic lipid metabolism disorders may exacerbate the disease and complicate management. With limited pharmacological options during pregnancy, plasmapheresis has emerged as a valuable therapeutic modality.

Case Presentation A 21-year-old primigravid woman at 31+1 weeks of gestation was admitted for follow-up of chronic pancreatitis and severe hypertriglyceridemia. Genetic testing confirmed a homozygous APO A5 mutation. Serial laboratory evaluations revealed triglyceride levels ranging from 600 to 1450 mg/dL, with associated anemia and fluctuating fibrinogen values. Plasmapheresis was initiated twice weekly. During follow-up, the patient required intermittent erythrocyte suspension and plasma replacement due to anemia and coagulopathy. Infection work-up was performed for recurrent fever episodes. Catheter-related infection was suspected; blood cultures grew Micrococcus luteus. Empiric ceftriaxone was initiated and discontinued after clinical improvement. Inflammatory markers normalized with therapy. Obstetric monitoring: Serial ultrasonography revealed fetal biometry consistent with gestational age. Amniotic fluid volume was adequate. Fetal cardiac activity and movements remained reassuring.

Discussion Pregnancy complicated by severe hypertriglyceridemia poses significant challenges, as standard lipid-lowering therapies are limited by teratogenic risk. In this patient, APO A5 homozygous mutation was the underlying cause, and plasmapheresis provided effective and safe lipid control throughout pregnancy.

CONCLUSION: This case demonstrates that in pregnancies complicated by APO A5 mutation–related hypertriglyceridemia and chronic pancreatitis, regular plasmapheresis is a feasible and effective therapy. Comprehensive multidisciplinary care and close obstetric surveillance are essential to optimize maternal and perinatal outcomes.

Keywords: Pregnancy, chronic pancreatitis, hypertriglyceridemia

SS-075

Bilateral Congenital Knee Dislocation in a Twin Pregnancy: A Case Report

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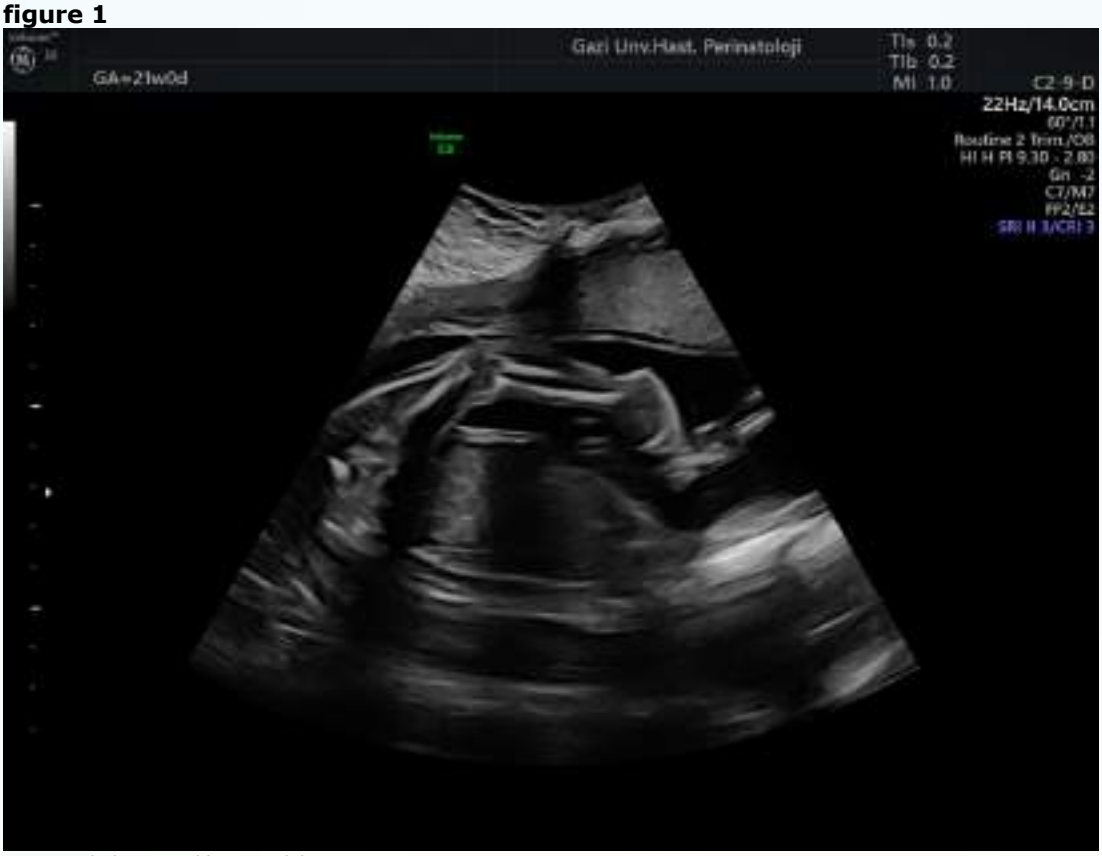
Congenital dislocation of the knee (CDK), or congenital genu recurvatum, is an extremely rare malformation (approximately 1 in 100,000 live births) characterized by hyperextension of the knee present at birth. The exact etiology of CDK is not completely understood, and many cases are considered idiopathic. It can occur in isolation or as part of a genetic syndrome. The presence of a persistent fetal knee hyperextension on serial scans should prompt detailed fetal anatomic survey for other anomalies and consideration of underlying syndromes. In such cases, prenatal genetic testing can be offered.

We report a case of a 34-year-old primigravida with dichorionic diamniotic twin gestation with in vitro fertilization (IVF). The anatomy screening at 20 weeks' gestation revealed an isolated knee hyperextension deformity in one twin (female fetus), raising suspicion for CDK. Amniocentesis for genetic evaluation was offered but declined. After birth it revealed that female neonate had bilateral knee hyperextension (genu recurvatum) with obvious knee joint dislocations. Bilateral developmental dysplasia of the hips (DDH) was confirmed postnatally by ultrasound. The infant underwent conservative orthopedic management after birth, but required bilateral knee joint reduction surgery with quadriceps tenotomy at 8 months.

This case highlights the rarity of bilateral CDK and its occurrence in a twin pregnancy. CDK can occur in the absence of known risk factors or syndromes, as a true idiopathic anomaly like. Early postnatal management with gentle reduction and casting is emphasized as the first-line treatment, with surgical intervention reserved for selected cases.

Keywords: Congenital knee dislocation, twin pregnancy, fetal limb dislocation

figure 1

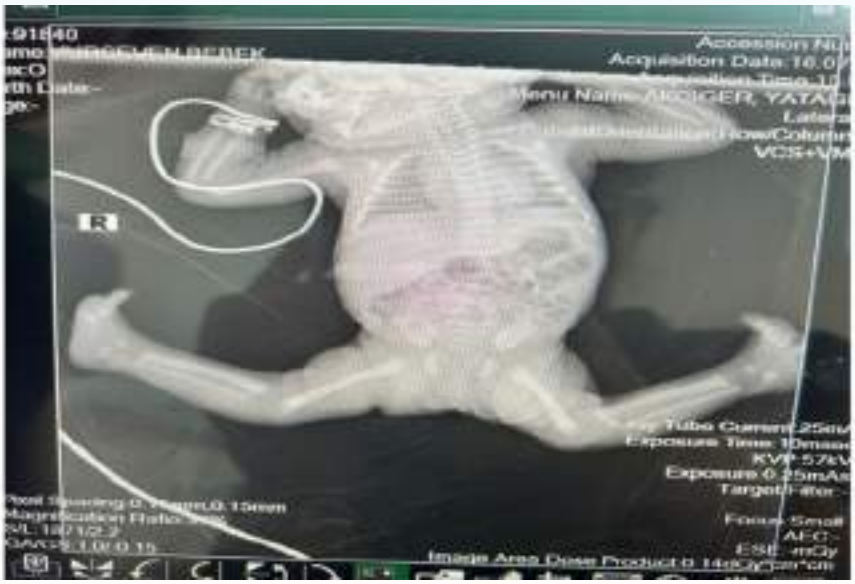


Antenatal ultrasound image of the case

Figure 2

Clinical appearance of the case in the postnatal period

Figure 3



postnatal x-Ray

SS-076

Gebelikte ve postpartum dönemde görülen Bell's paralizisi:Olgu serisi

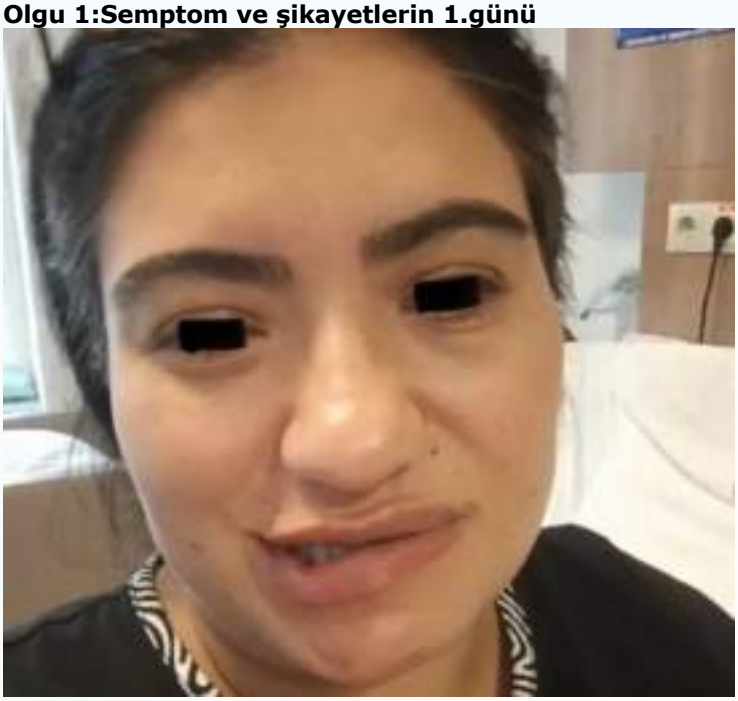
Dilek Tektaş, Şule Birol İnce, Petek Feriha Uzuner
Şişli Hamidiye Etfal Eğitim ve Araştırma Hastanesi,Kadın Hastalıkları ve Doğum Ana Bilim Dalı,İstanbul

Olgu 1:Gebeliğinin 31.haftasında olan hasta,kliniğimizde pregestasyonel diyabet,preeklampsi tanılarıyla takip ediliyordu.Takibi yapılırken sağ gözünü tam kapatamama,sol ağız köşesinde çekilme ve konuşmada zorluk gibi şikayetler tarifleyen hasta Nöroloji,Enfeksiyon,Kulak Burun Boğaz Hastalıkları kliniğine danışıldı.Nöroloji kliniği diffüzyon MR görülmesi ve Göz Hastalıkları kliniğine danışılmasını önerdi.Görüntülemede patoloji saptanmadı.Kulak Burun Boğaz Hastalıkları tarafından değerlendirilen hastada sağ grade 4 fasial paralizi bulguları saptandı.Tek doz 1 mg/kg metilprednizolon tedavisi önerildi.Enfeksiyon Hastalıkları kliniğine danışılan hastanın kapsamlı tetkikleri istendi.Herpes Simpleks Virüs(HSV) IgG pozitif,IgM negatif izlenen hastaya antiviral tedavi önerilmedi.Göz Hastalıkları kliniğine danışılan hastaya sodyum karboksimetilselülöz içeren göz damlası verildi.Tek doz metilprednizolon tedavisi aldıktan sonra hastanın sağ kulakta şişlik şikayeti üzerine Kulak Burun Boğaz Hastalıkları kliniğine danışıldı.Sağ grade 6 fasial paralizi tespit edildi.Metilprednizolon tedavisi düzenlenmiştir.Tedavisini tamamlayan hasta 35.gebelik haftasında ağrılı eski sezaryen tanısıyla sezaryene alındı ve canlı bir erkek bebek doğurtuldu.Takiplerinde genel durumu iyi, vitalleri stabil izlenen hasta poliklinik kontrolü önerileriyle taburcu edildi.

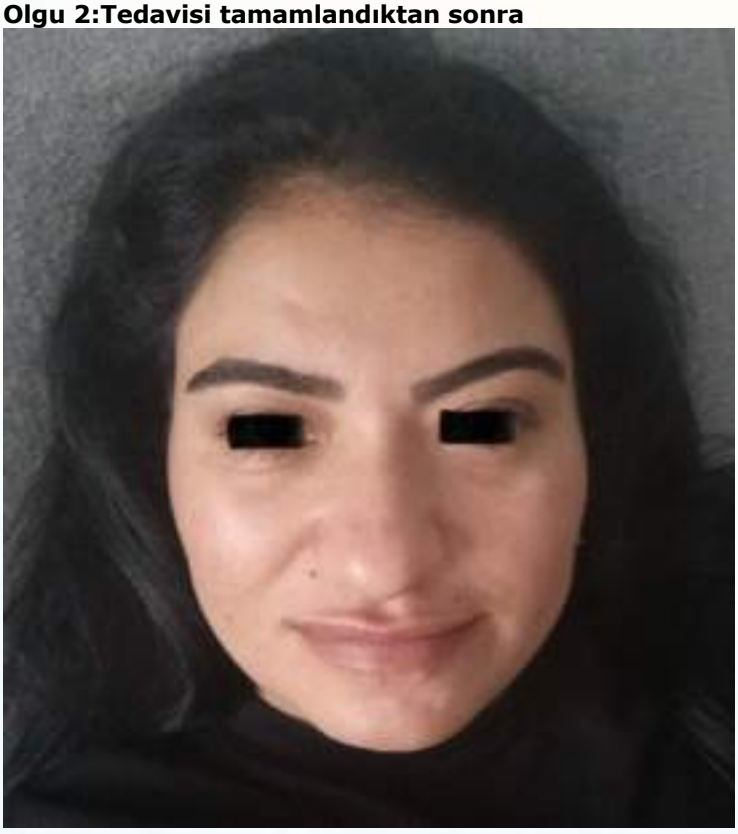
Olgu 2: Gebeliğinin 37. haftasında olan hasta,kliniğimizde preeklampsi,Tip 2 Diyabetus Mellitus tanılarıyla takip ediliyordu.Takibi yapılırken HELLP endikasyonuyla sezaryene alındı ve canlı bir erkek bebek doğurtuldu.4 yıl önce periferik fasyal paralizisi geçiren hastanın postpartum şikayetleri başlayınca Nöroloji,Göz ve Kulak Burun Boğaz Hastalıkları kliniğine danışıldı.Göz Hastalıkları kliniği tarafından değerlendirilen hastada oftalmolojik patoloji izlenmedi.Nöroloji kliniği tarafından değerlendirilen hasta muayenesinde nistagmusu olduğu görülmüştür.Sol göz sıkma gücü sağa göre daha zayıf,sol ağız köşesi sağa göre düşük izlendi.Bu belirtiler Bell’s paralizisi sekeli olarak değerlendirildi.Hastanın kranial görüntülemesinde patolojik bulgu izlenmedi.Paralizi sekeli dışında muayenesi doğal görüldü.Paralizi sekelinin metilprednizolon tedavisine yanıt vermeyeceği düşünüldü.Genel durumu stabil olan hasta kontrol önerileriyle taburcu edildi.

Anahtar Kelimeler: periferik fasyal paralizi, preeklampsi,metilprednizolon

Olgu 1:Semptom ve şikayetlerin 1.günü



Olgu 2:Tedavisi tamamlandıktan sonra



SS-077

Prenatal Diagnosis of Extrapulmonary Sequestration: Report of Two Cases

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Ankara Etlik City Hospital Gynecology and Obstetrics Clinic Perinatology Department

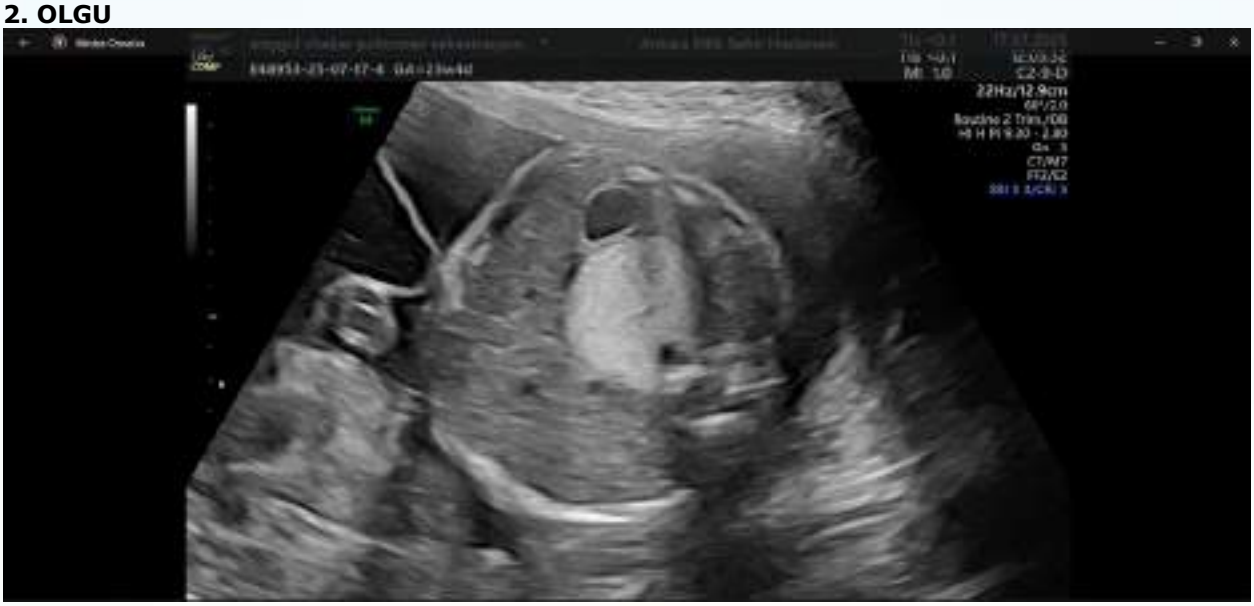
OBJECTIVE: Extrapulmonary sequestration (EPS) is a rare congenital lung malformation. The objective of this study was to present two cases of prenatally diagnosed EPS and to emphasise the diagnostic contribution of ultrasonography and complementary imaging.

METHODS-RESULTS: The study evaluated two pregnant women. In the initial case, a 28-year-old G2A1, a 15-mm hyperechogenic mass, was identified at the left renal–suprarenal junction. Color Doppler imaging revealed systemic arterial supply from the aorta, and fetal magnetic resonance imaging was scheduled for further assessment. The second case study concerned a 42-year-old female patient who was undergoing in-vitro fertilisation treatment for a twin pregnancy. A 31-mm mass was observed extending from the left lung into the subdiaphragmatic region, resulting in anterior displacement of the stomach. Doppler imaging was utilised to confirm the presence of vascular supply from the aorta.

CONCLUSION: The prenatal diagnosis of EPS is feasible through ultrasonography, with Doppler evaluation of the systemic vascular supply being critical for confirmation. Whilst the prognosis is generally favourable, the size of the lesion, its location and associated anomalies have a significant influence on clinical outcomes. The differential diagnosis encompasses a range of potential diagnoses, including neuroblastoma, renal or suprarenal cysts, congenital cystic adenomatoid malformation, and congenital lobar emphysema. The cases presented herein illustrate that EPS can be reliably identified in utero, thus highlighting the need for multidisciplinary prenatal counselling and tailored perinatal management.

Keywords: : Extrapulmonary Sequestration, Prenatal Diagnosis, Ultrasonography, Doppler, Fetal MRI

2. OLGU



31-mm mass was observed extending from the left lung into the subdiaphragmatic region, resulting in anterior displacement of the stomach. Doppler imaging was utilised to confirm the presence of vascular supply from the aorta.

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SS-078

Right Aortic Arch with Right Ductus Arteriosus: CASE REPORT

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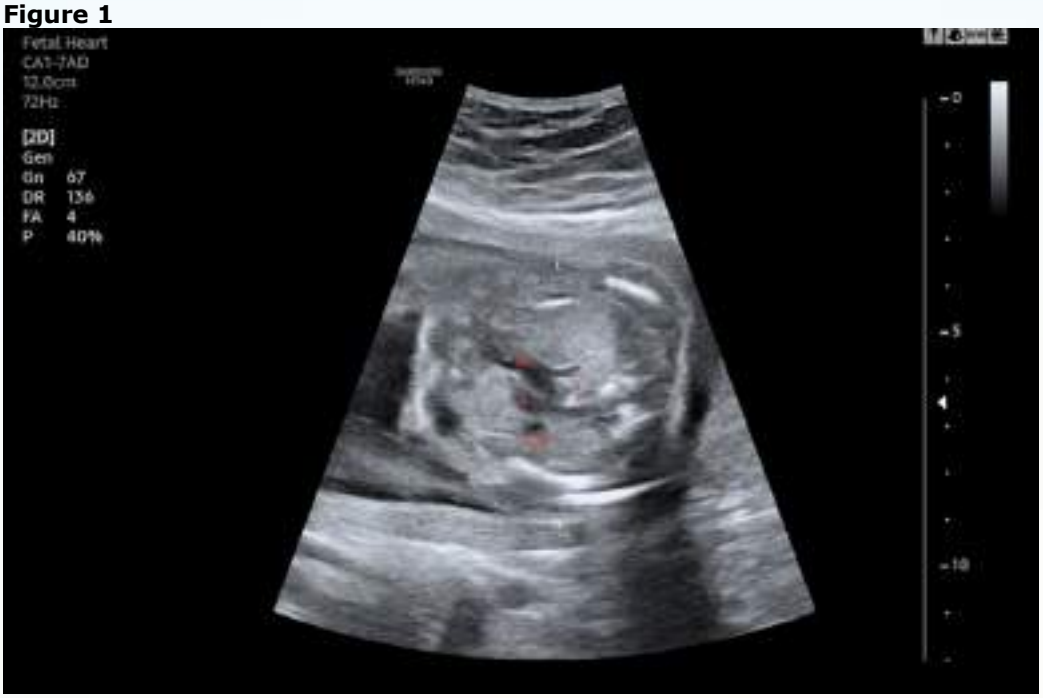
INTRODUCTION: Right aortic arch(RAA) diagnosed on fetal echocardiography when the transverse arch is located to the right of the trachea. In cases with a right ductus arteriosus(RDA), both vessels lie on the right side, and the aorta and pulmonary arteries converge in a 'V-shaped' pattern in right side of the trachea without creating a vascular ring.[1]

CASE REPORT: A 33-year-old patient, G3P2C/S2Y2, was referred to our clinic at 24 weeks of gestation after detailed anatomical ultrasonography revealed a RAA and single umblical artery(SUA). Fetal echocardiography confirmed a RAA with a RDA. Amniocentesis with karyotyping and chromosomal microarray at 25 weeks were normal. The pregnancy follow-up was uneventful, female infant with apgar 4/7 was delivered by cesarean at 38 weeks of gestation. At 6 months of age, the infant remained asymptomatic and has continued routine cardiac follow-up.

DISCUSSION: Vascular rings are aort arch anomalies that may range from incidentally detected asymptomatic lesions to those causing tracheoesophageal compression. RAA with a RDA is typically isolated and doesn't influence prognosis in the absence of other cardiac defects.[1] Although an isolated SUA isn't regarded as a risk factor for chromosomal abnormalities, the concurrent identification of a RAA anomaly in our case warranted genetic evaluation with karyotyping and microarray analysis.[2] Despite the absence of chromosomal abnormalities, the coexistence of these findings proved important guidance during prenatal follow-up. Prenatal diagnosis of aortic arch anomalies is important, as it enables timely surgical intervention, reduces postoperative residual symptoms, and provides prognostic value through the assesment of associated anomalies.[3]

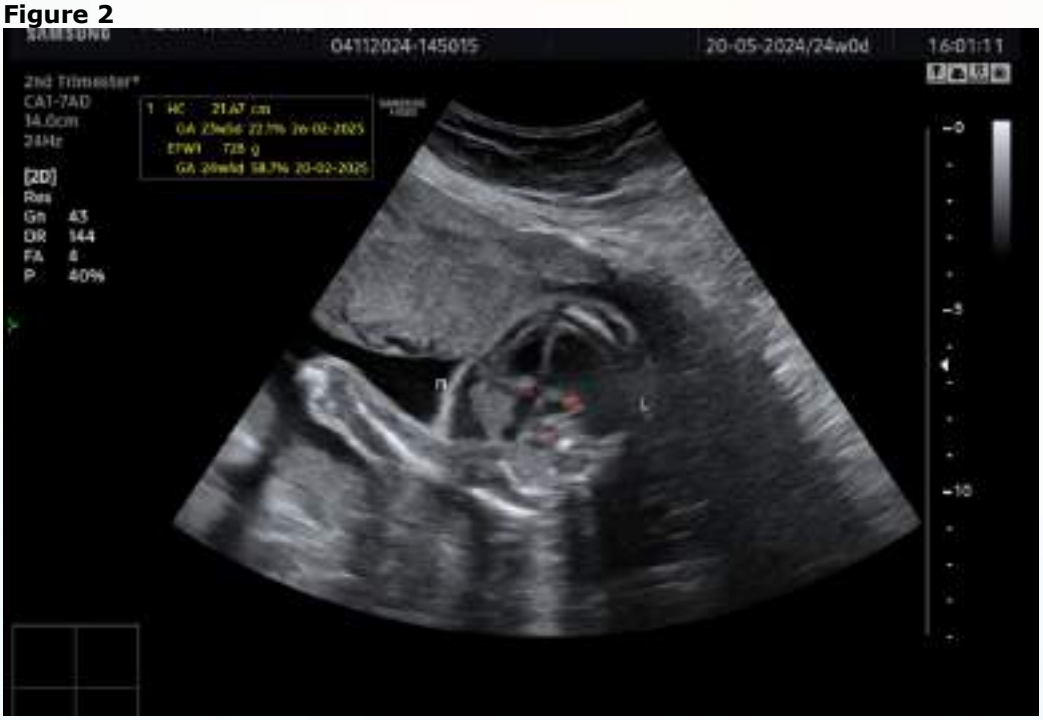
Keywords: right aortic arch, right ductus arteriosus, single umblical artery, vascular rings

Figure 1



Right-sided aortic arch and right-sided ductus arteriosus with the 'V-shaped' image observed on the right side of the trachea. PA: pulmonar artery, A: aorta, SVC: superior vena cava, T: trachea

Figure 2



Pulmonar artery and aorta observed on the right side of the trachea PA: pulmonar artery, A: aorta, T: trachea

POSTER BILDIRILER

PS-001

The Diagnostic Pitfall of Low Risk NIPT in the Face of Evolving Fetal Anomalies: A DiGeorge Syndrome Case Report

Aliye Erdogan, Sezin Uludağ, Tuğçe Arslanoğlu, Oğuzhan Yürük, Alev Atış Aydın, Deniz Kanber Acar
Kanuni Sultan Süleyman Eğitim ve Araştırma Hastanesi, Kadın Hastalıkları ve Doğum Ana Bilim Dalı, Perinatoloji Bölümü

INTRODUCTION: DiGeorge Syndrome is a significant genetic disorder associated with a spectrum of clinical manifestations, notably conotruncal cardiac defects and immunodeficiency. Tetralogy of Fallot is one of the most frequently observed cardiac anomalies in DiGeorge syndrome. We present a challenging case where a fetus, initially monitored due to an isolated finding of nasal bone agenesis in the first trimester, was ultimately diagnosed with DiGeorge Syndrome after multiple severe anomalies were detected later in pregnancy.

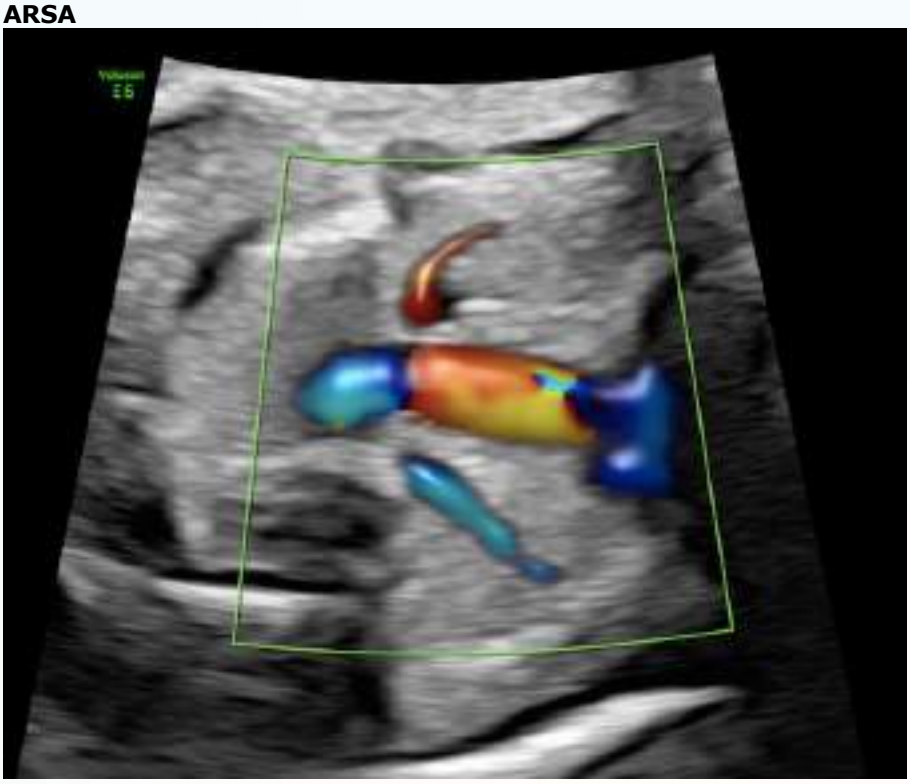
Case Presentation A 23 year old, G1P0 patient presented at 11 weeks and 4 days for first-trimester screening. Ultrasound revealed a normal NT but an absent nasal bone. The patient was counseled on invasive testing but opted for NIPT.

At 22 weeks and 4 days, detailed ultrasound revealed multiple severe findings; pulmonary artery stenosis, ventricular septal defect, overriding aorta, ARSA and right renal agenesis was also detected. Despite the NIPT returning a low risk result, the presence of these complex anatomical anomalies mandated amniocentesis. FISH and array analyses confirmed the diagnosis of DiGeorge Syndrome, after which the patient elected for termination following detailed counseling.

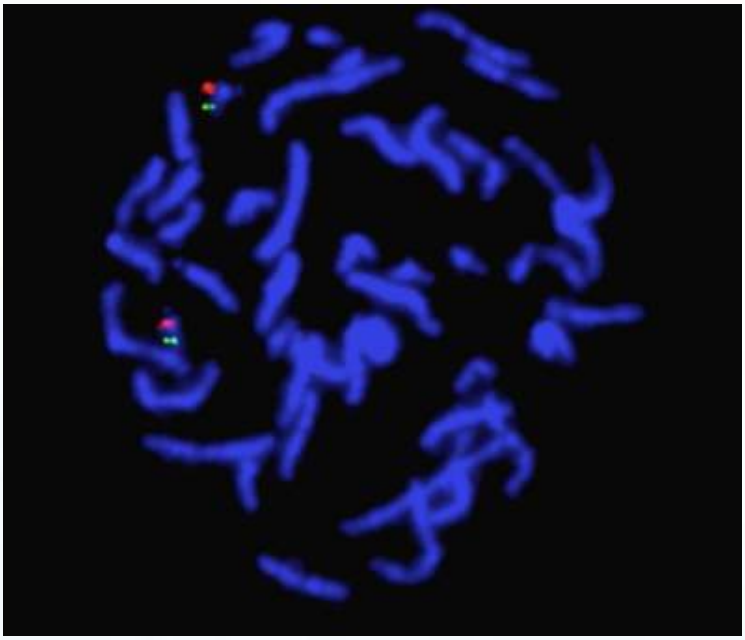
CONCLUSION: This case underscores that an isolated early ultrasound marker can predate the emergence of complex fetal anomalies in later gestation. Crucially, it highlights that a low risk NIPT result does not eliminate the necessity for definitive invasive diagnostic procedures when serious anatomical defects are identified by ultrasound. The case emphasizes the paramount importance of prioritizing ultrasound findings in fetal surveillance and the critical role of comprehensive genetic counselling.

Anahtar Kelimeler: DiGeorge Syndrome, Fetal Cardiac Anomalies, Genetic Diagnosis,

ARSA

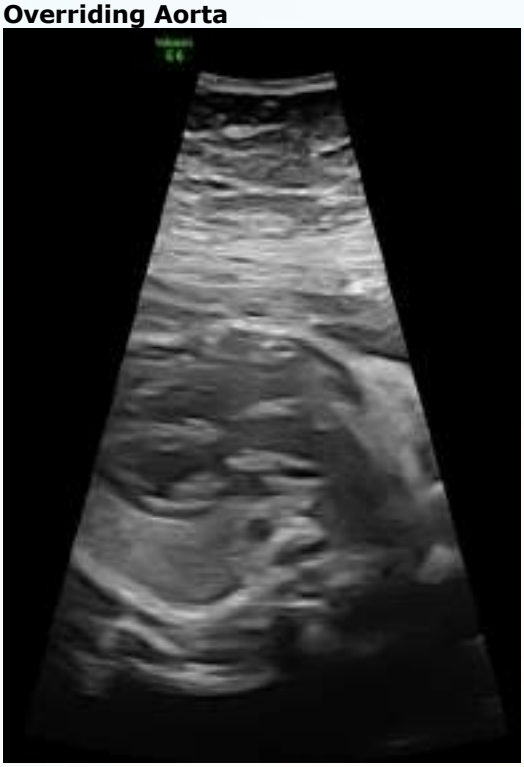


FISH

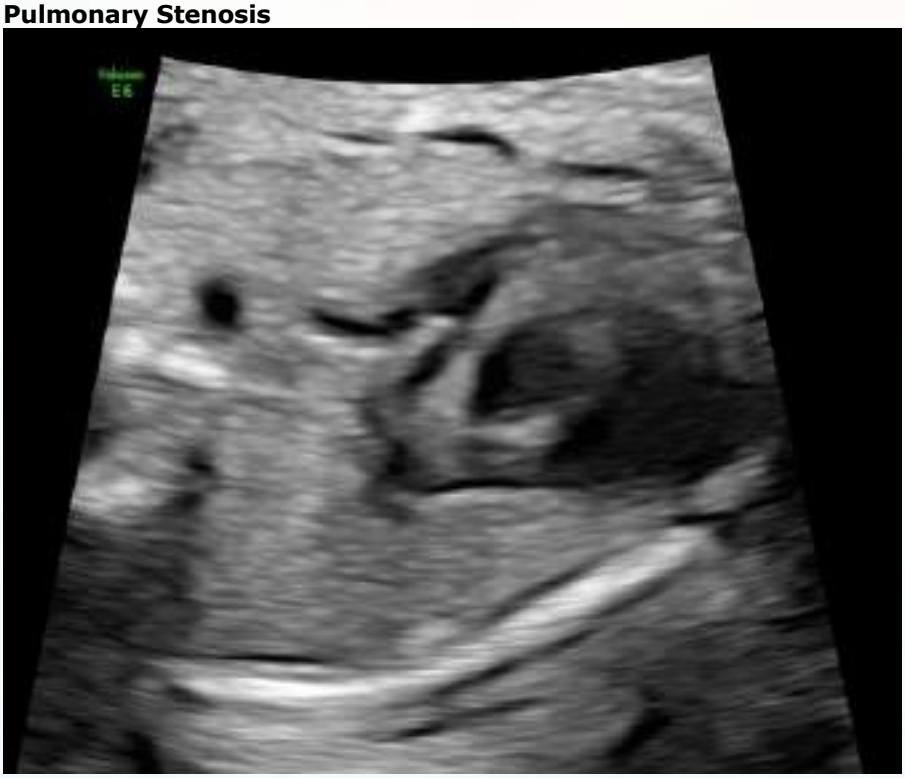


DiGeorge "TUPLE1" (22q11.2) / 22q13.3 (SHANK3) probe Cytocell Cell count: 100

Overriding Aorta



Pulmonary Stenosis



PS-002

Turner Syndrome Diagnosed by Multicystic, Septated Cystic Hygroma in an Adolescent Pregnancy: A Case Report

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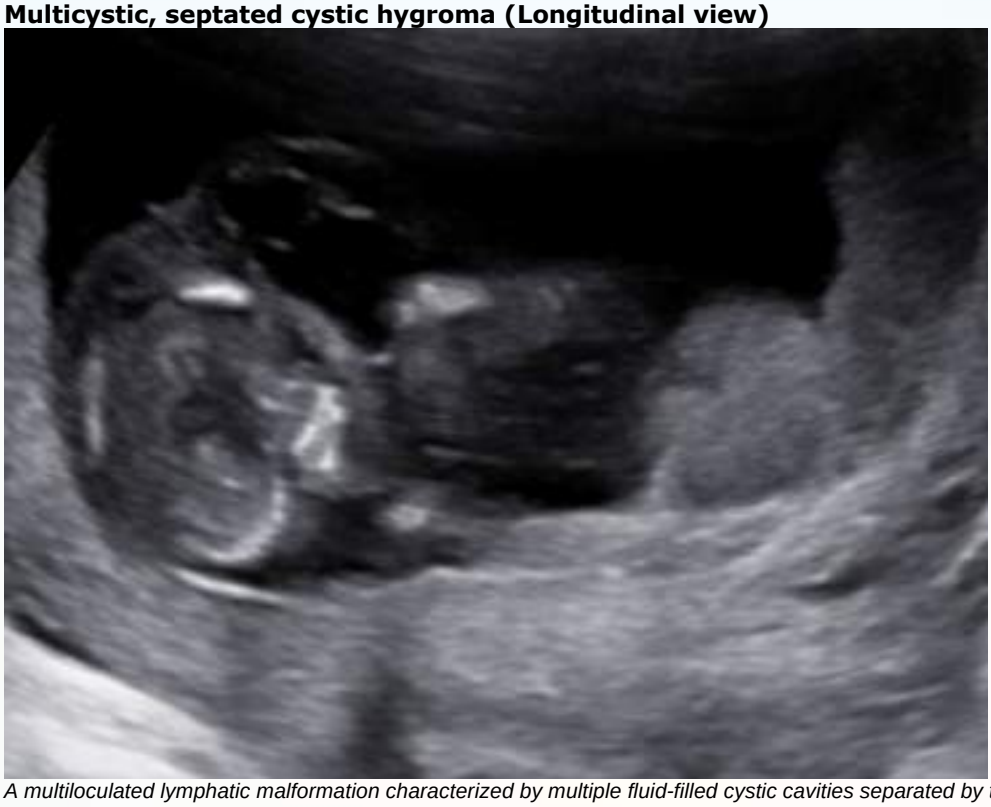
INTRODUCTION: Turner syndrome, caused by complete or partial monosomy X, is one of the most frequent chromosomal abnormalities detected in early pregnancy. Characteristic first-trimester sonographic findings include cystic hygroma, especially with septations, which strongly correlate with adverse outcomes. Early detection provides critical information for prognosis and counseling. CASE: A 15-year-old primigravida presented at 13 weeks + 5 days of gestation. First-trimester ultrasound demonstrated a multicystic, septated cystic hygroma in the cervicothoracic region, consisting of multiple fluid-filled cavities separated by thin septa. No additional structural malformations were observed. Chorionic villus sampling was performed, and cytogenetic analysis confirmed Turner syndrome (45,X). At follow-up, absent fetal cardiac activity was identified. A medical termination of pregnancy was subsequently performed according to clinical protocols. DISCUSSION: The presence of a multicystic, septated cystic hygroma in the first trimester is a hallmark ultrasonographic marker of chromosomal anomalies, most commonly Turner syndrome. This lymphatic malformation results from impaired jugular lymphatic sac development and inadequate venous drainage, producing multiloculated cystic structures. Prognosis is poor, with most cases leading to miscarriage, intrauterine fetal demise, or severe neonatal morbidity. Although spontaneous regression has occasionally been reported, survival in monosomy X remains rare. This case illustrates the classical presentation and natural course of Turner syndrome, emphasizing the role of early targeted sonography and confirmatory invasive testing for diagnosis and counseling, particularly in adolescent pregnancies. CONCLUSION: Early recognition of Turner syndrome by ultrasound and genetic testing enables timely diagnosis, appropriate management, and essential counseling for patients and families.

Keywords: multicystic, septated cystic hygroma; adolescent pregnancy, Turner syndrome

Multicystic, septated cystic hygroma (Longitudinal view)



A multiloculated lymphatic malformation characterized by multiple fluid-filled cystic cavities separated by thin septations, located in the cervicothoracic region.

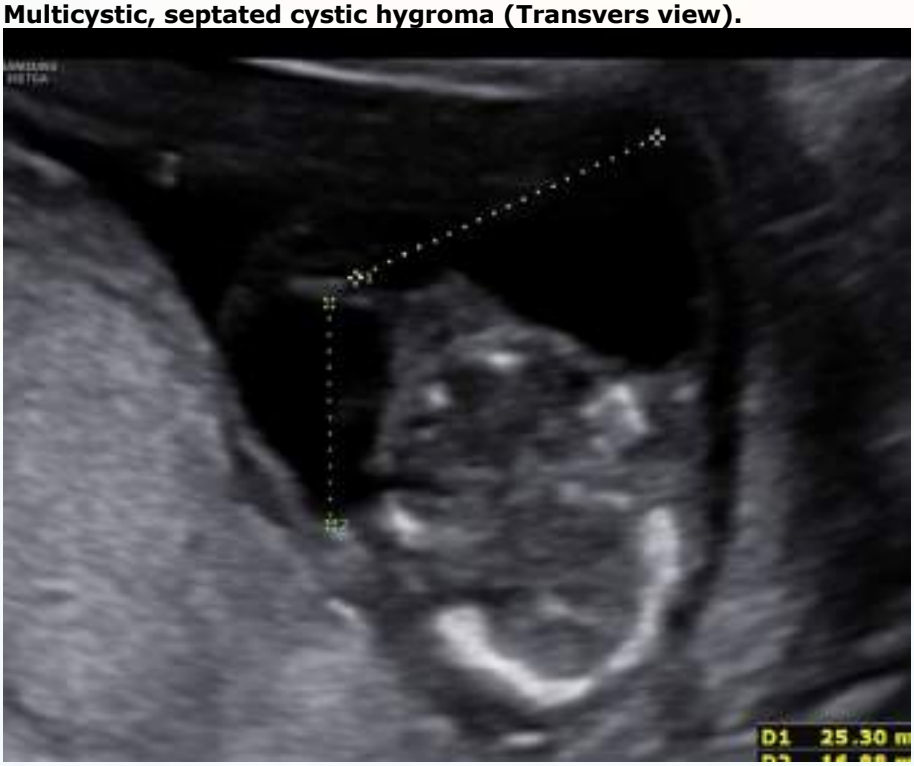
Fetal Conditions Associated with Septated Cystic Hygroma.

Category	Fetal / Genetic Condition	Description	Approximate Frequency
Chromosomal abnormalities	Turner syndrome (45,X)	Most common chromosomal abnormality associated with septated cystic hygroma due to lymphatic drainage obstruction.	~20–40%
	Trisomy 21 (Down syndrome)	May present with nuchal edema and septations; frequently associated with congenital heart defects.	~10–15%
	Trisomy 18 (Edwards syndrome)	Commonly associated with multisystem malformations; poor prognosis.	~5–10%
	Trisomy 13 (Patau syndrome)	Often accompanied by craniofacial and cardiac anomalies.	~2–5%
	Triploidy (69,XXX, etc.)	Typically results in early pregnancy loss.	<2%
Microdeletion / duplication syndromes	22q11.2 deletion / duplication	Associated with conotruncal heart defects and craniofacial dysmorphism.	Rare

Monogenic / syndromic causes	Noonan syndrome (RASopathy)	Septated cystic hygroma may occur in euploid fetuses with PTPN11, SOS1, or RAF1 mutations.	~5–10% (among euploid cases)
	Fryns, Neu-Laxova, and Multiple Pterygium syndromes	Inherited genetic disorders, usually lethal.	Very rare
Structural anomalies	Congenital heart defects	Frequently associated with AVSD, Tetralogy of Fallot, or other major cardiac malformations.	~60–70% (in normal karyotype fetuses)
	Skeletal and CNS anomalies	Includes hydrocephalus, arthrogryposis, agenesis of corpus callosum, and scoliosis.	Variable
Fetal circulation / hydrops	Non-immune fetal hydrops	Results from lymphatic obstruction, cardiac failure, or chromosomal defects.	Commonly associated
Transient / benign causes	Transient lymphatic obstruction	May regress spontaneously in early gestation; usually benign course.	Rare (~10%)

The presence of septations indicates a significantly higher risk of aneuploidy compared to isolated increased nuchal translucency. In euploid fetuses, further testing (WES, CMA, or PTPN11 gene analysis) should be considered to evaluate for RASopathies. Prognosis depends on karyotype, presence of hydrops, and associated anomalies; mortality exceeds 80% in Turner and trisomy cases.

Multicystic, septated cystic hygroma (Transvers view).



A multiloculated lymphatic malformation characterized by multiple fluid-filled cystic cavities separated by thin septations, located in the cervicothoracic region.

PS-004

A Case Report of 17q12 Deletion Diagnosed Prenatally

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Division of Perinatology, Department of Obstetrics and Gynecology, Gazi University Faculty of Medicine, Ankara, Turkey

BACKGROUND: 17q12 microdeletion syndrome is a rare genetic disorder caused by the loss of a segment in the long arm of chromosome 17 (region q12). It is associated with a broad phenotypic spectrum including renal malformations, neurodevelopmental delay, intellectual disability, and endocrine dysfunction. Although most cases occur de-novo, genetic counseling is essential due to its variable presentation and potential familial occurrence.

Case Presentation: A 29-year-old gravida 2 para 1 woman, with no history of consanguinity, presented at 23 weeks of gestation. Her family history was notable for Alström syndrome. Prenatal ultrasound revealed bilateral hyperechogenic kidneys and increased nuchal fold thickness (7 mm). Amniocentesis confirmed a 17q12 microdeletion. At 37 + 6 weeks, spontaneous rupture of membranes led to vaginal delivery of a female infant weighing 2510 g with Apgar scores of 8 and 9 at 1 and 5 minutes, respectively. The newborn, now four month old, is under multidisciplinary follow-up.

CONCLUSION: Prenatal detection of bilateral renal hyperechogenicity and increased nuchal fold thickness should prompt consideration of chromosomal abnormalities, including 17q12 microdeletion. Given the risk for renal dysfunction, developmental delay, and endocrine disorders such as maturity-onset diabetes of the young (MODY), comprehensive postnatal evaluation and multidisciplinary management are essential. This case highlights the importance of targeted genetic testing for accurate diagnosis and anticipatory care planning.

Keywords: 17q12 microdeletion, renal hyperechogenicity, prenatal diagnosis, chromosomal deletion, MODY

PS-005

A Rare Case of Congenital High Airway Obstruction Syndrome (CHAOS) Diagnosed in the Second Trimester

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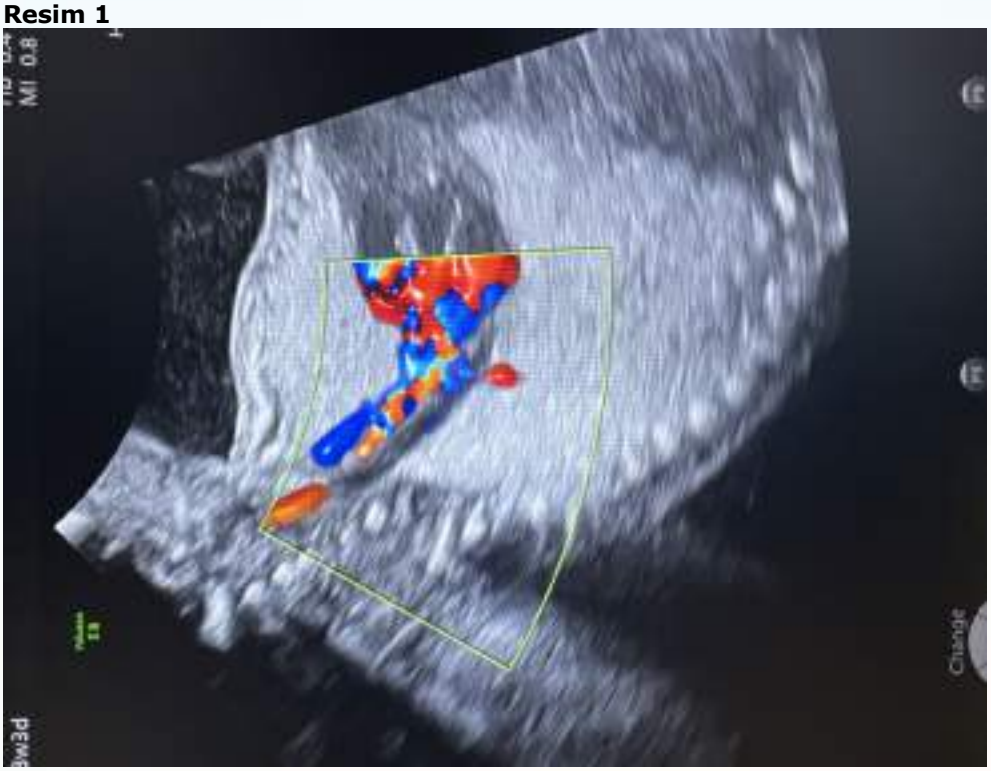
BACKGROUND: Congenital High Airway Obstruction Syndrome (CHAOS) is a rare and often fatal condition resulting from partial or complete obstruction of the fetal upper airway, most commonly due to laryngeal or tracheal atresia or stenosis. Sonographic features typically include bilaterally enlarged hyperechoic lungs, dilated airways, flattened or inverted diaphragm, ascites, and a compressed heart. Secondary findings may include fetal hydrops and polyhydramnios due to esophageal compression. CHAOS may be associated with chromosomal anomalies such as trisomy 9, trisomy 16, and 5p deletion, or may occur as part of Fraser syndrome. The estimated incidence is approximately 1 in 50,000 pregnancies.

Case Presentation: We present the case of a 25-year-old primigravida with no previous surgical history and a background of hypothyroidism. First-trimester ultrasound and screening results were unremarkable. At 18 weeks and 3 days of gestation, routine second-trimester ultrasonography revealed bilateral echogenic enlarged lungs, inverted diaphragms, decreased cardiothoracic ratio,hyperecogenic bowel,intracardiac echogen focus and ascites. These findings were consistent with CHAOS. Amniocentesis was performed, and FISH analysis showed a normal disomic pattern.

CONCLUSION: This case highlights the importance of early recognition of CHAOS during routine prenatal screening. Although rare, CHAOS carries a poor prognosis due to severe cardiopulmonary compromise. Early diagnosis enables timely counseling regarding prognosis and management options, including the possibility of pregnancy termination or consideration of fetoscopic and EXIT procedures in selected cases.

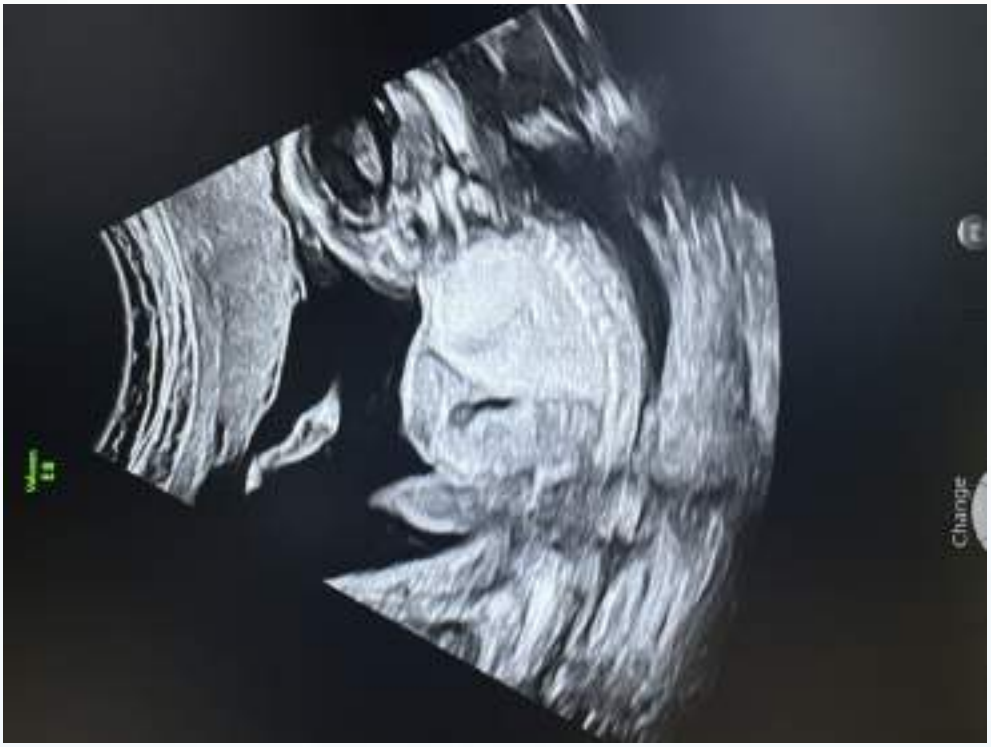
Keywords: Congenital High Airway Obstruction Syndrome (CHAOS),hyperechoic lung,inverted diaphragm

Resim 1



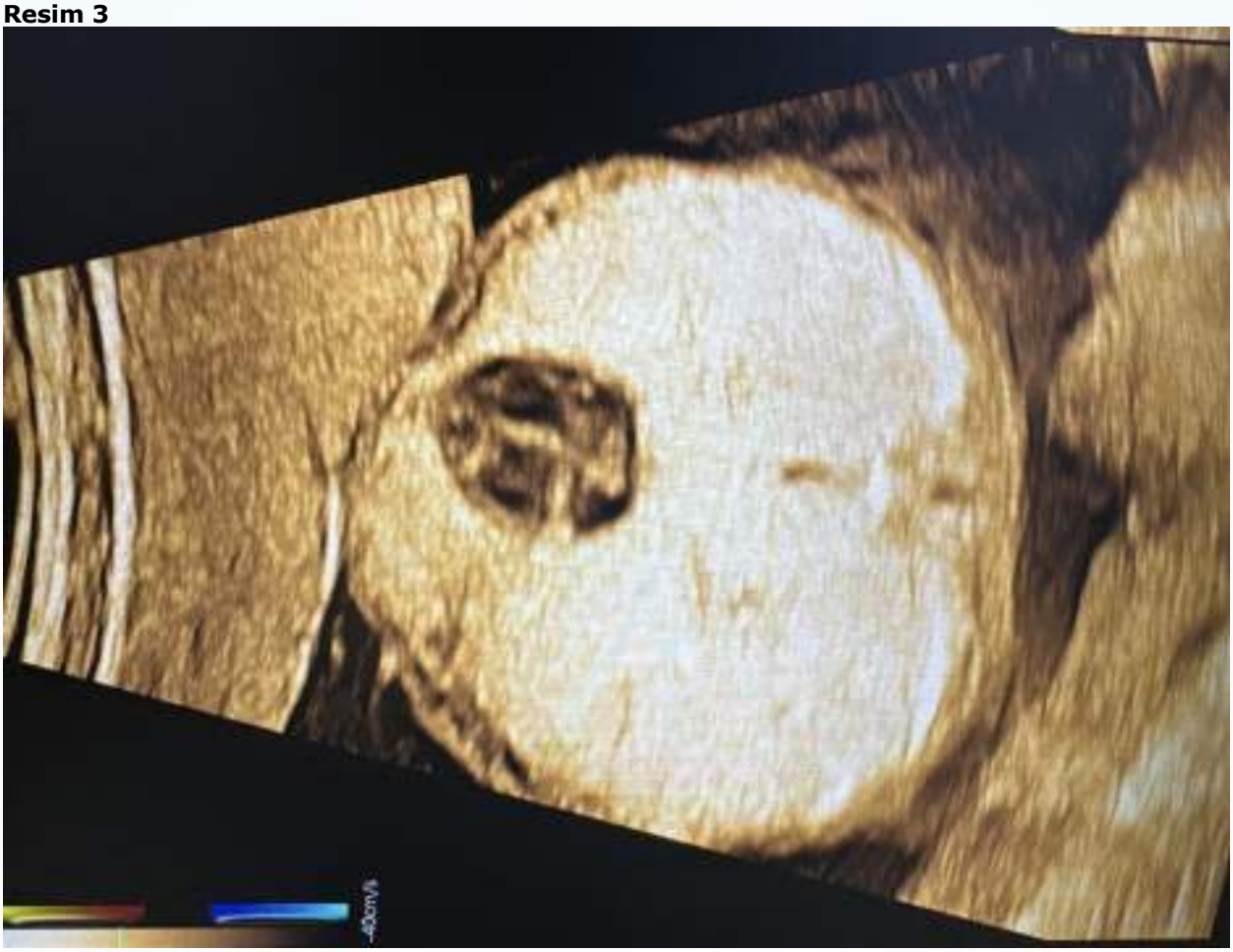
Damar posteriorunda dilate havayolu izlenmekte

Resim 2



Akciğer genişlemiş ve hiperekojen görünümde, diafragma batına doğru everte görünümde izlendi.

Resim 3



Kardiotorasik oran azalmış, kalp ortada komprese olmuş, akciğerler hiperekojen izlendi.

PS-006

Redundant Foramen Ovale Flap Presenting with Fetal Arrhythmia: A Case Report

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Redundant foramen ovale flap (RFOF) is usually considered a benign anatomical variant; however, in certain cases it may act as a substrate for atrial irritability and clinically significant arrhythmias. We present a case of RFOF initially detected due to fetal arrhythmia.

A 28-year-old gravida 3 para 2 was referred at 32+3 weeks of gestation for fetal bradycardia. Ultrasonography revealed polyhydramnios, a large-for-gestational-age fetus, and 2:1 atrioventricular block. The foramen ovale flap extended to the left ventricular wall with an opening of 8.3 mm, while other cardiac structures were normal. Fetal echocardiography demonstrated premature atrial contractions. During follow-up, supraventricular tachycardia developed, with rates up to 270 bpm. Digoxin therapy was initiated, subsequently escalated, and sotalol was added. After rhythm control was achieved, sotalol was discontinued, and the patient was discharged at 36 weeks under digoxin therapy. At 37+4 weeks, the patient underwent cesarean delivery. A 3045 g male neonate with APGAR scores of 8 and 10 was delivered. The newborn required three days of intensive care. Propranolol (1 mg/kg/day) was initiated and continued after discharge.

In conclusion, when detected prenatally, RFOF should not only be regarded as an incidental finding but also as a potential focus of arrhythmogenesis. This case illustrates that RFOF may present with both bradyarrhythmia and supraventricular tachycardia. As supported by current literature, digoxin remains the first-line therapy, while sotalol or flecainide may be considered in refractory cases. Careful echocardiographic surveillance, multidisciplinary perinatal management, and individualized therapy enabled favorable outcomes in both prenatal and postnatal periods.

Keywords: fetal arrhythmia, fetal arrhythmia therapy, redundant foramen ovale flap

PS-007

Rare Intestinal Atresias: Two Case Reports

Hatice Aydın Salman, Arzu Sümer Ortaç, Esra Nuhoğlu Bilgin, Esin Şahin Toruk, Bilal Emre Erzeneoğlu, Pınar Tokdemir Çalış, Tuncay Nas, Mehmet Zeki Taner, Deniz Karçaaltıncaba
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In duodenal atresia,the tract between the proximal and distal portions of the duodenum is atretic. Incidence is 1/2500–1/10000 live births.Ultrasound diagnosis:Double bubble, with communication between the two parts; late polyhydramnios.Risk of aneuploidy is high (20–50%),mainly trisomy 21. Jejunal atresia incidence is 1/2500–1/5000 live births.Ultrasound diagnosis:Severe late-onset dilation of the loops proximal to the obstruction(which is absent in most cases prior to 25 weeks of gestation), late-onset polyhydramnios.Risk of aneuploidy is low.

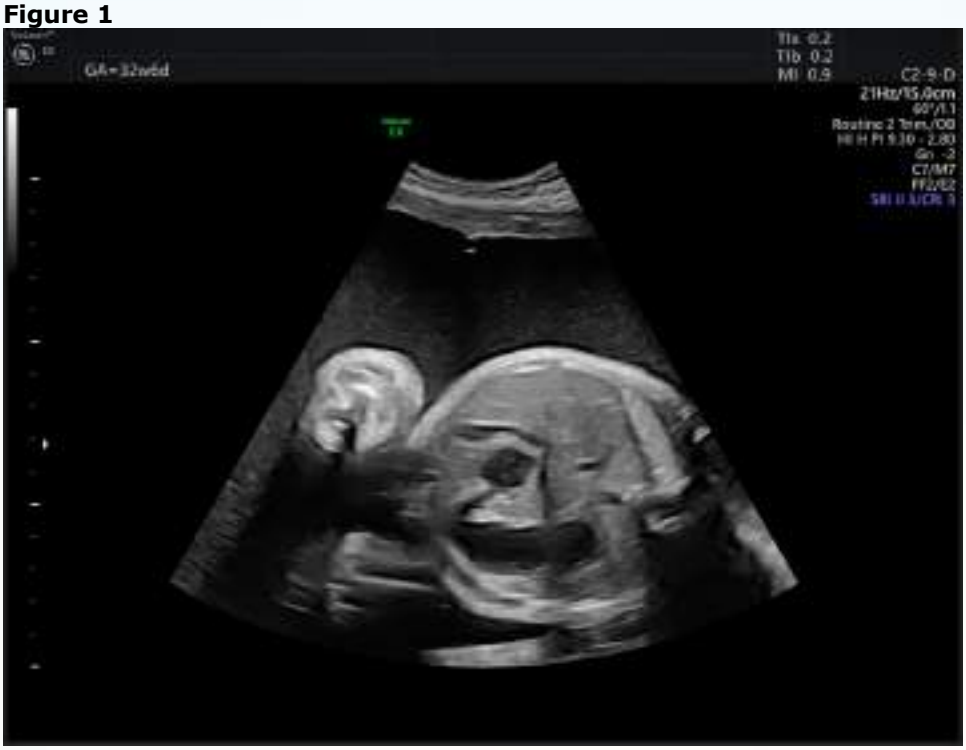
Case 1: 30-year-old,gravida 1,IVF pregnancy. At 27 weeks of gestation,ultrasonography revealed a double bubble sign and polyhydramnios.Annular pancreas was suspected.Amniocentesis was performed and both karyotype and array analyses were found to be normal.A female infant with FGR(2275 g, Apgar 8/9)was delivered by cesarean section at 37 weeks.Postpartum,duodenal atresia and annular pancreas were identified in the infant.Surgical intervention was performed,consisting of Kimura's diamond-shaped-duodenoduodenostomy.

Case 2: 27-year-old,gravida 1. At 30 weeks of gestation, ultrasonography revealed polyhydramnios and dilated bowel loops. At 33 weeks, following preterm prelabor rupture of membranes(pprom)and onset of preterm labor,a male infant(1805 g,Apgar 7/9) was delivered vaginally.Following birth,jejunal atresia and short bowel were diagnosed and surgical intervention was performed(jejunojejunal anastomosis, jejunostomy,serial transverse enteroplasty)

In duodenal atresia,outcome is mainly good if isolated.But more severe anomalies of the biliary tract and of the pancreas (annular pancreas),which may have a negative impact on prognosis. In jejunal atresia,the risks of preterm birth and fetal growth restriction (FGR) are increased.Outcome is generally good but is poor in multiple-site atresia.

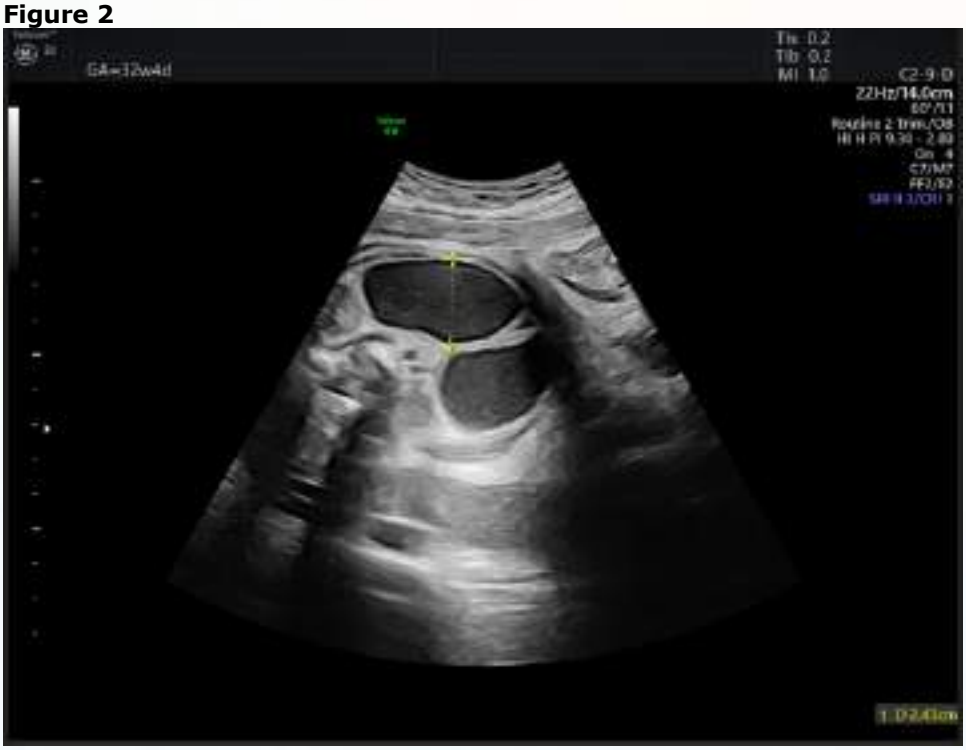
Keywords: Annular pancreas,jejunal atresia,duodenal atresia

Figure 1



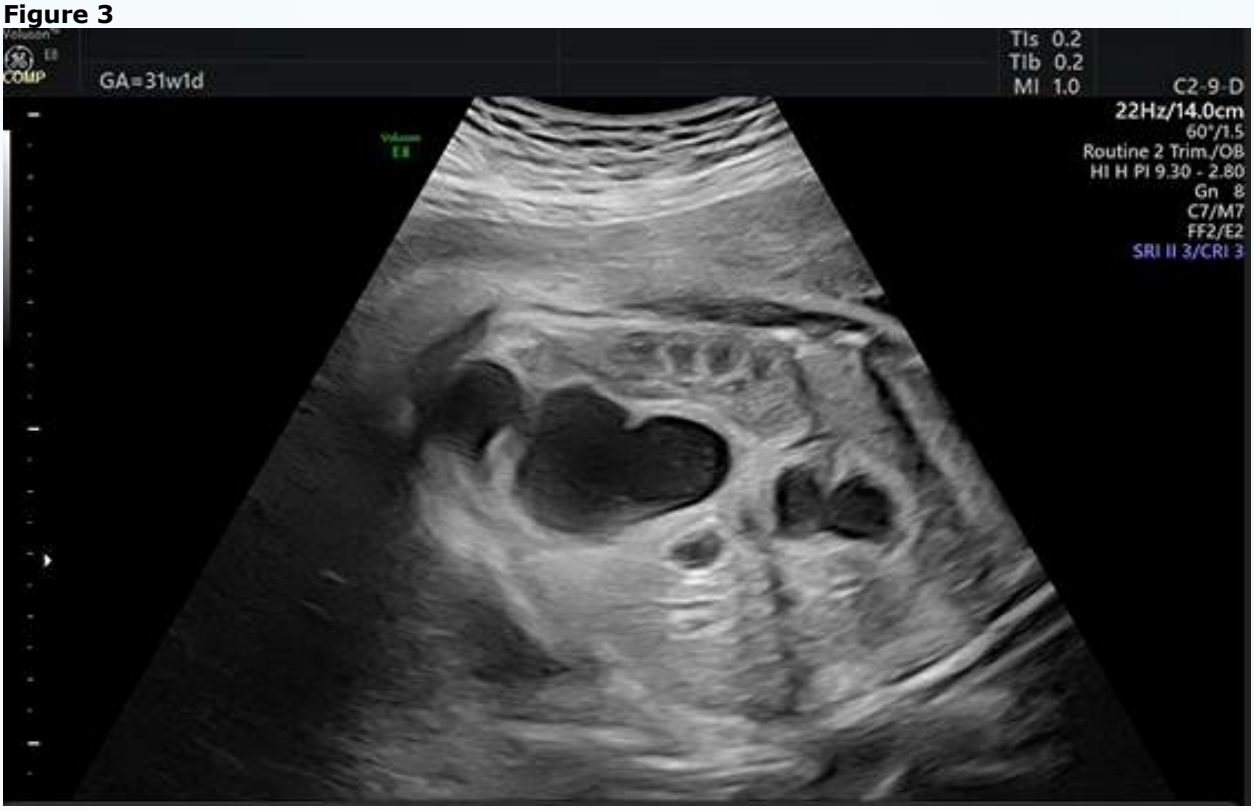
Annular pancreas.

Figure 2



Intestinal dilatation, jejunal atresia

Figure 3



Jejunal atresia

Türkiye Maternal Fetal Tıp ve Perinatoloji Derneği
Ultrasonografi Kongresi
29 Ekim - 1 Kasım 2025
Renaissance Polat İstanbul Hotel - Yeşilköy

PS-008

Prenatal Diagnosis of Snyder–Robinson Syndrome (SMS Gene) Following Detection of Short Long Bones and Dilated Fetal Bowel Loops

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İntroduction: Snyder–Robinson syndrome (SRS) is an ultra-rare X-linked intellectual disability syndrome due to loss-of-function variants in SMS. Reported features include characteristic facial dysmorphism, asthenic build with low muscle mass (often within the first year of life), progressive kyphoscoliosis, early-onset osteoporosis with atraumatic fractures, and seizures. Developmental delay typically presents as failure to meet early milestones and evolves to mild-to-profound intellectual disability; motor disability is variable but cognitive impairment appears relatively stable over time.

CASE: A 37-year-old pregnant woman, gravida 4, parity 1, abort 2, was referred to our clinic at the 23rd week of gestation due to do a detailed ultrasonography in the fetus. The patient was not history of a consanguineous marriage. In the current pregnancy, ultrasonography showed short femur length (FL) and short humeral length (HL) relative to gestational age, with dilated fetal bowel loops. Amniocentesis was performed. Fetal genetic testing identified a hemizygous pathogenic variant in SMS, consistent with Snyder–Robinson syndrome. Maternal whole-exome sequencing (WES) detected no pathogenic variant, supporting a likely de novo origin of the fetal variant. Findings and implications (X-linked inheritance, phenotypic spectrum, postnatal needs, and options for pregnancy management) were discussed in a multidisciplinary setting. The family opted for termination of the pregnancy.

CONCLUSION: This case illustrates that standard cytogenetic analysis and chromosomal microarray may be insufficient when prenatal ultrasound shows short long bones with additional anomalies. Early incorporation of exome sequencing into the prenatal work-up can yield a timely, actionable diagnosis, such as SRS, and inform counseling and management..

Keywords: Prenatal Diagnosis,SMS gene, Snyder Robinson Syndrome,

PS-009

Crohn hastalığı olan gebede apendektomi sonrası maternal sepsis ve preterm doğum olgusu

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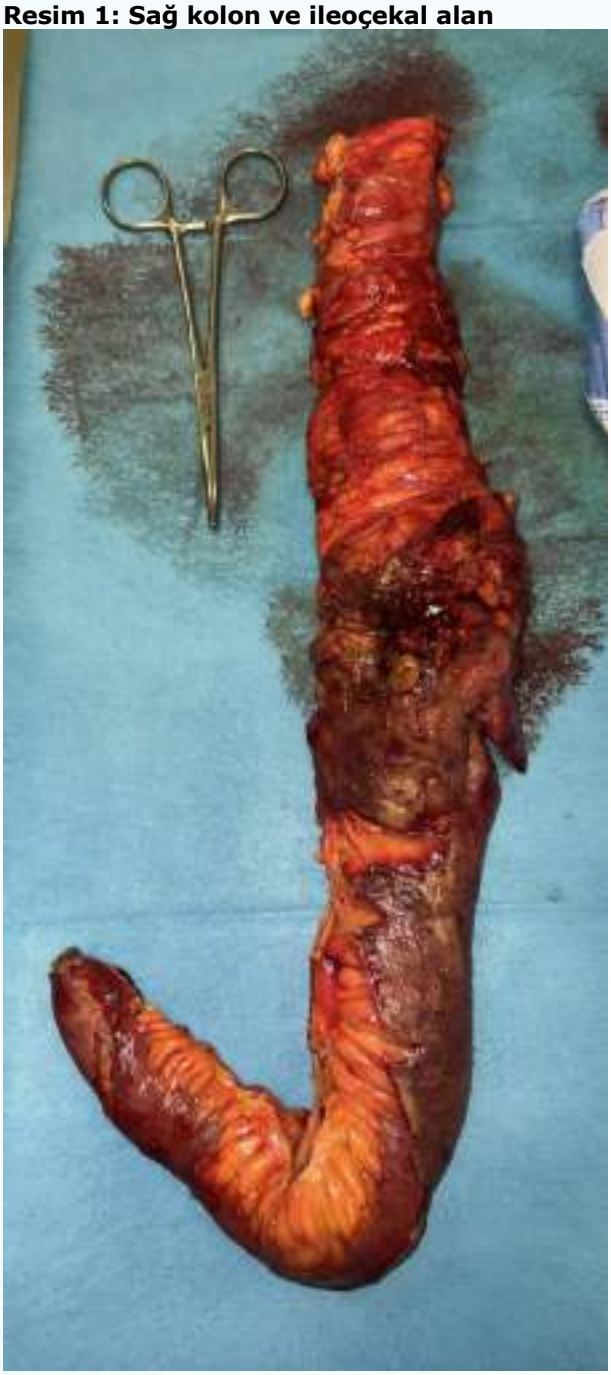
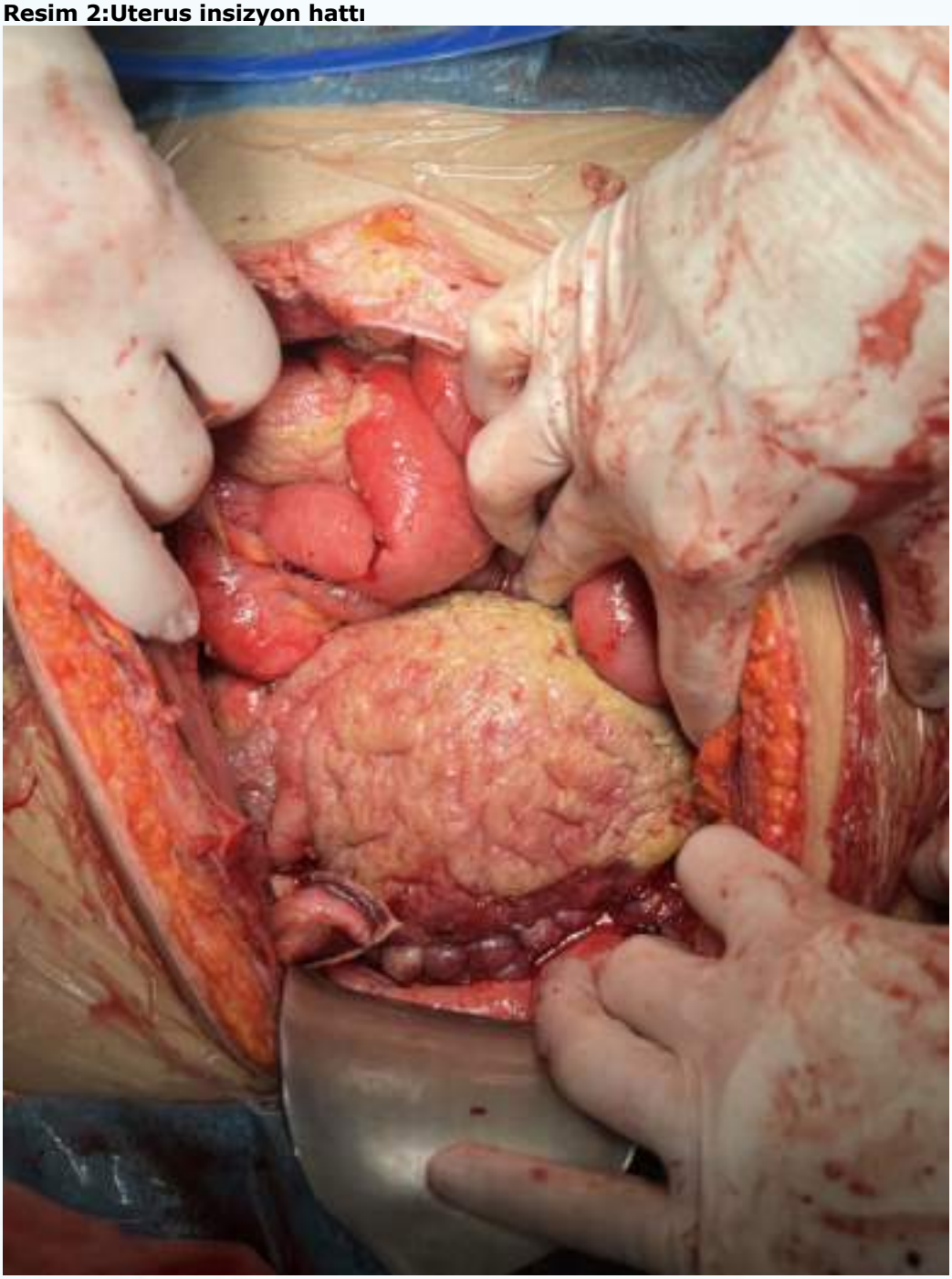
30 yaşında gebeliğinin 27.haftasında olan hasta karın ağrısı şikayetiyle başvurduğu genel cerrahi kliniğinde takip ediliyordu.Hastanın antenatal tarama testlerinden fetal DNA analizi düşük risk olduğu görüldü.Crohn hastalığı tanısı olan gebe azatioprin ve adalimumab tedavisi almaktadır. Akut apandisit şüphesiyle istenen MRI görüntülemede apendiks çapı 6,7 mm ve periapendiküler doku inflame izlendi.Yapılan ultrasonografide apendiks çapı 8,1 mm izlendi ve Crohn hastalığına bağlı sekonder apandisit ile uyumlu izlendi.Tarafımızca değerlendirilen hastanın yapılan ultrasonografide fetal biyometri ölçümleri haftasıyla uyumlu,serviks uzunluğu 35 mm izlendi ve uterin kontraksiyon görülmedi. Maternal taşikardi ve CRP değerinin 164 görülmesi üzerine enfeksiyon hastalıkları tarafından değerlendirildi.Kan kültürleri istenen hastaya günlük seftriakson 2 gr,metronidazol 500 mg 8 saatte 1 verilmek üzere başlandı.Tarafımızca değerlendirilen hastanın akciğer matürasyonu açısından betametazon dozlarının 24 saat arayla tamamlanması,nöroprotektif etki amacıyla magnezyum tedavisi başlanması ve sonrasında operasyona alınması önerildi.Ateşi yükselen ve şikayetleri artan hasta genel cerrahi ve kadın hastalıkları ve doğum tarafından operasyona alındı.Akut batın tablosuyla operasyona alınan hastanın perfore apandisit olduğu görüldü ve apendektomi yapıldı.Peroperatif ultrasonografi ile tarafımızca intrauterin fetal kalp atımı pozitif olduğu görülerek operasyon sonlandırıldı.Hasta 27 hafta 3 günlük iken postoperatif 1.gününde magnezyum tedavisi 24 saat tamamlanınca sonlandırıldı ve betametazon dozları tamamlandı.Postoperatif 7.gününde dreninde bağırsak içeriği izlenen ve sepsis tablosu gelişen gebe maternal sepsis endikasyonuyla 28 hafta 2 günlük iken sezaryene alındı.Genel cerrahi tarafından değerlendirilen iskemik alanlar nedeniyle sağ kolon ve ileoçekal rezeksiyon yapılmasına karar verilerek sağ hemikolektomi yapıldı.Apgar skoru 1.dakika 4,5.dakika 6 olan 31 cm 1145 gram canlı kız bebek doğurtuldu.Hasta postoperatif genel yoğun bakıma devredildi.Postoperatif 7.gününde erken komplikasyon görülmeyen hasta genel cerrahi kliniği tarafından devralındı.

Anahtar Kelimeler: Crohn hastalığı,preterm eylem,sepsis

Resim 1: Sağ kolon ve ileoçekal alan



Resim 2:Uterus insizyon hattı



PS-010

Prenatal Fallot Tetralojisi ve Doğum Sonrası Kardiyak Bulgular

Zeynep Balsu, Şule Birol İnce, Feriha Petek Uzuner
Şişli Hamidiye Etfal Hastanesi, Kadın Hastalıkları ve Doğum Kliniği

GİRİŞ: Fallot Tetralojisi,Ventriküler Septal Defekt, ata binen aorta, sağ ventrikül çıkış yolu obstrüksiyonu ve ikincil kardiyomegali ile tanımlanan konotrunkal bir anomalidir.Prenatal tanı perinatal yönetimi belirler.

OLGU: 29 yaşında G1P0 gebe;SAT 27 Ekim,ilk trimester kombine testi düşük risk. 19+3'te ayrıntılı değerlendirmede fetal ekokardiyografide kardiyomegali,kalp aksında sola deviasyon,interventriküler septumda defekt, ata binen aorta, pulmoner çıkış yolunda stenoz ve üç damar-trakea kesitinde aort çapının pulmoner arterden büyük olduğu (aorta>PA) saptandı;tek umbilikal arter izlendi. 21+3'te yinelenen fetal ekoda aynı bulgular(VSD, overriding aorta, pulmoner stenoz, aorta>PA, kardiyomegali,sola aks) doğrulandı ve ToF ile uyumlu kabul edildi.

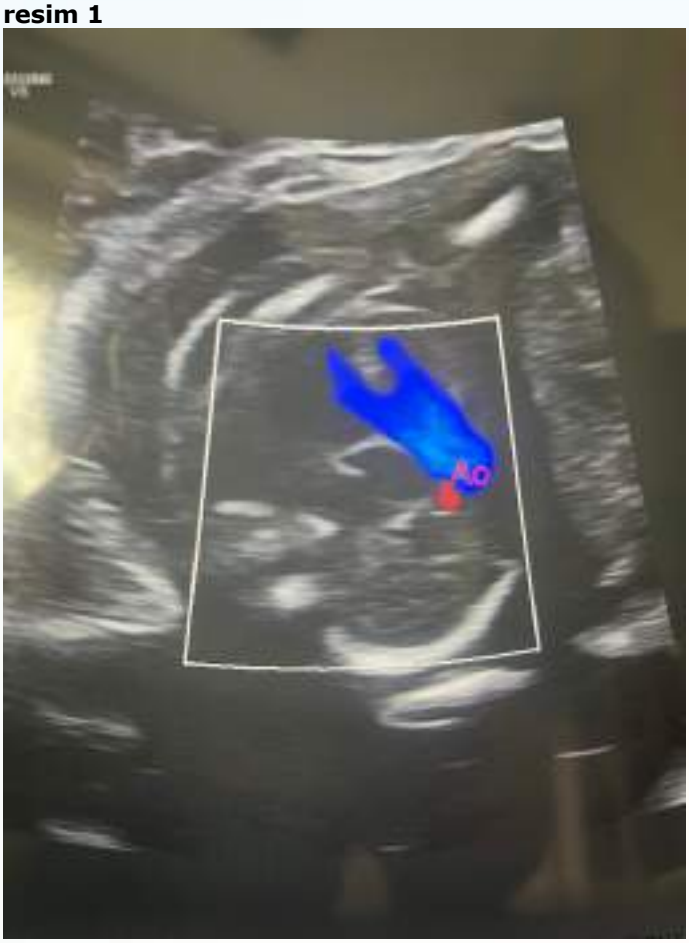
Doğum sonrası değerlendirme:Postnatal ekokardiyografide kardiyak apeks solda, situs solitus, atriyoventriküler bağlantılar konkordant, aort-pulmoner arter ilişkisi normal bildirilmiştir.Ventriküler loop D-loop pulmoner ve sistemik venöz dönüşler normal;atriyal septum intakt. Mitral ve triküspid kapaklar yapısal olarak normal,TY ile sağ ventrikül basıncı 18 mmHg. Muskuler ventriküler septumda defekt izlenmemiştir.Aort arkusu/istmus normal;koarktasyon yok.Duktus arteriozus kapanmış,perikard/endokard/myokard patolojisi saptanmamıştır. Ölçümlerde aort %80 dekstropoze, pulmoner gradiyent 52 mmHg,infrarenal (inen) aort gradiyenti 17 mmHg, aortik gradiyent 12 mmHg,pulmoner arter çapı 3,8 mm olarak bildirilmiştir.

TARTIŞMA: Postnatal yaklaşım doğumdan sonra da anlaşılabilecek anatomik farklılıklara bağlıdır.Pulmoner arter ve dallarındaki darlığın derecesi,multiple VSD varlığı ve koroner arter anomalilerinin varlığı bu farklılıklardandır ve her biri tedavi şekline yön verir.Multidisipliner izlem, seri ekokardiyografi, genetik değerlendirme ve ek anomaliler açısından fetal anatomi taraması gereklidir.

SONUÇ: Prenatal ayrıntılı kardiyak tarama ve 3VT kesiti, konotrunkal anomalilerin erken tanısında kritiktir;postnatal tekrar değerlendirme tanının spektrum içindeki konumunu netleştirir ve perinatal yönetimi yönlendirir.Bizim hastamız şu anda 3 aylık günde 5mg propranolol kullanıyor.Pediatrik kardiyoloji tarafından aylık kontrollerine gidiyor.Opere olması için uygun vücut ağırlığına gelmesi bekleniyor.

Anahtar Kelimeler: Fallot Tetralojisi, Fetal ekokardiyografi, Konjenital Kalp Hastalığı

resim 1



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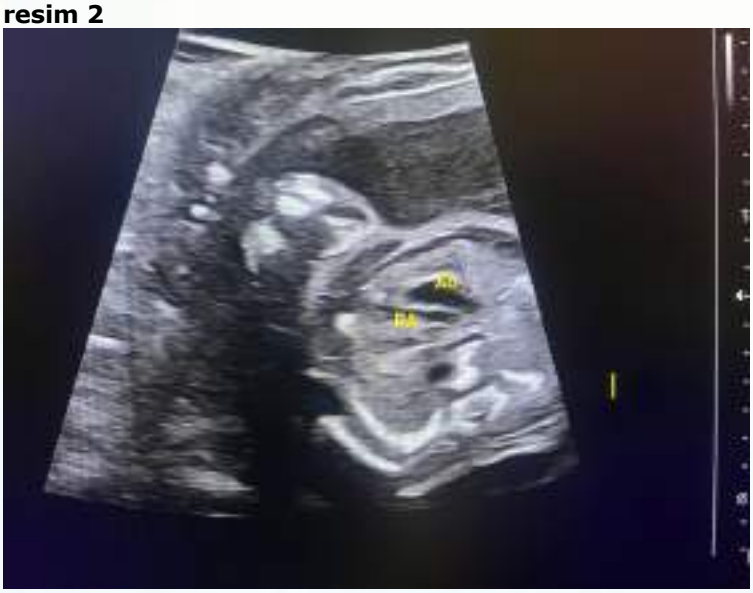

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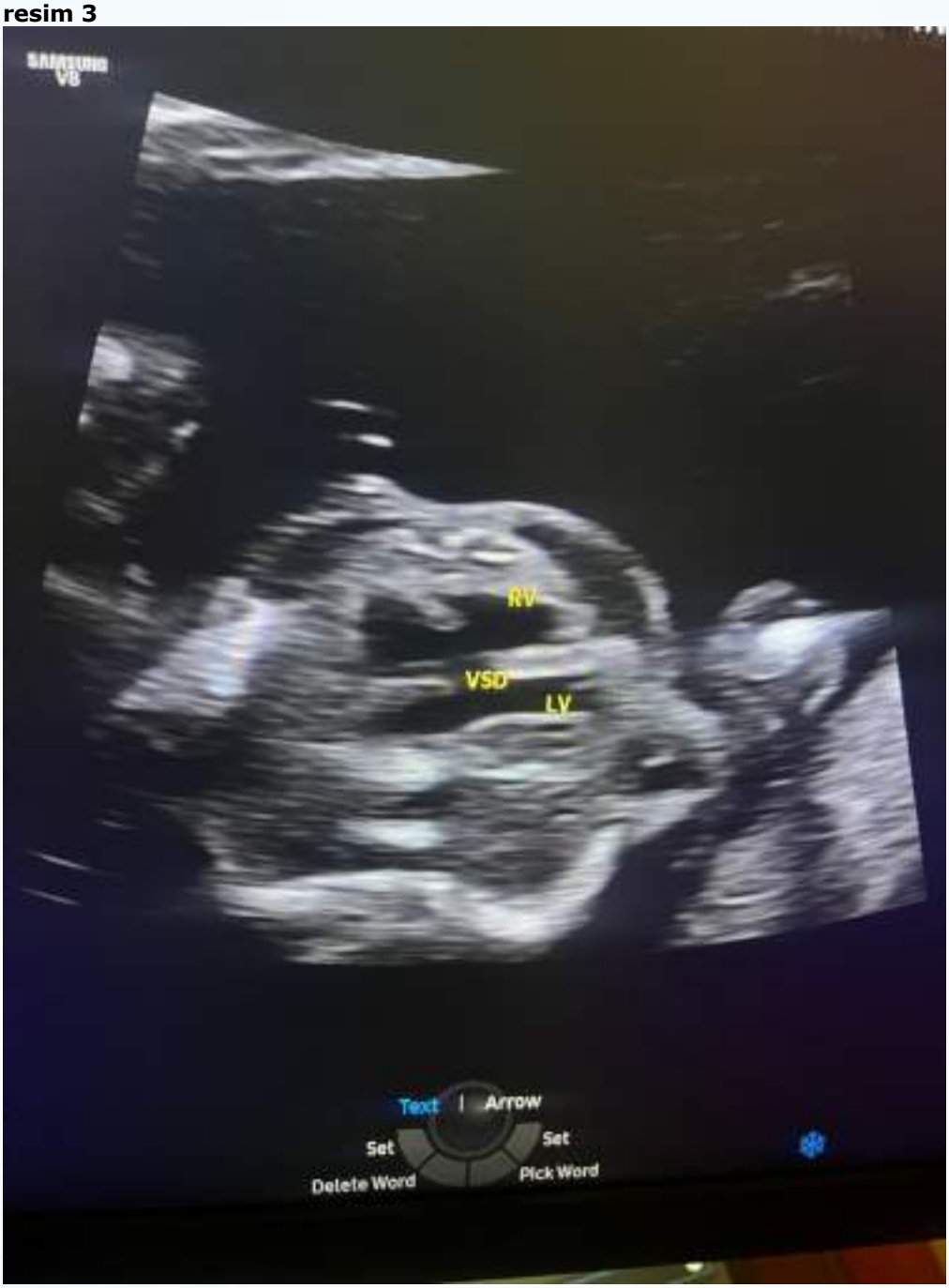

PS-011

A case of multiple intraabdominal cysts detected in the fetus at 14 weeks of gestation and diagnosed postnatally as *lymphangioma*

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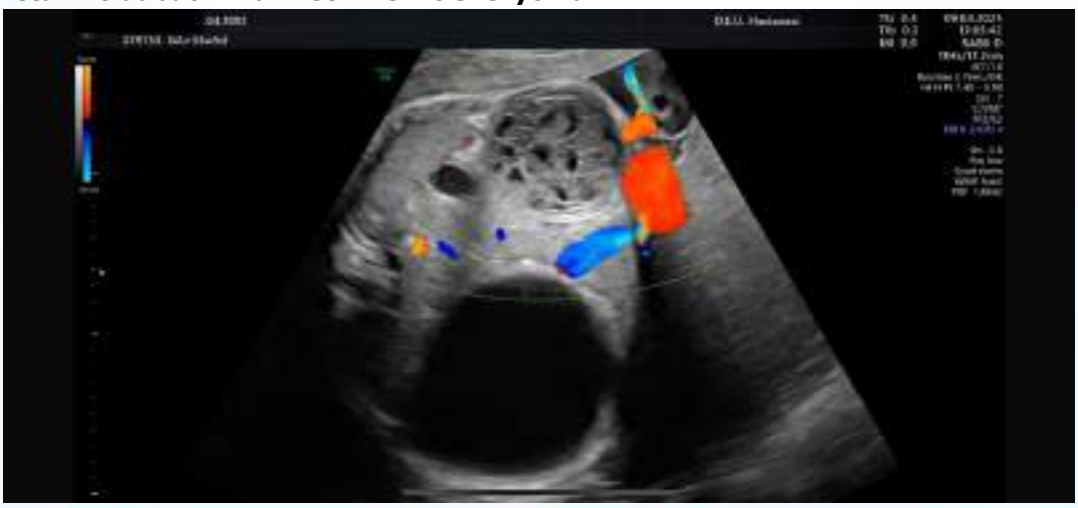
OBJECTIVE: Fetal intraabdominal cystic lesions are rare and heterogeneous in origin. They may be associated with mesenteric cysts, gastrointestinal duplication cysts, ovarian cysts, or lymphangioma. Prenatal recognition and differentiation are crucial for prognosis and planning perinatal management. We present a case of prenatally detected intraabdominal cysts later diagnosed as lymphangioma.

CASE: A 32-year-old woman, G5P3A1, underwent ultrasound at 14+5 weeks of gestation, which revealed three cystic lesions in the fetal abdomen. During follow-up, one of these cysts collapsed and disappeared in the subsequent weeks. Amniocentesis showed normal results. At 25 weeks, fetal MRI and ultrasound confirmed two thin-walled cystic lesions measuring 7×8×8.5 cm on the right and 4×4×5 cm on the left, with septations in the right cyst. Cyst drainage with cytological evaluation was suggested but declined by the patient. During follow-up, fetal growth restriction developed. At 34+5 weeks, spontaneous vaginal delivery occurred. The female neonate weighed 2400 g, with Apgar scores of 7 and 9, and was admitted to the neonatal intensive care unit. On the 23rd postnatal day, surgical excision of the mass was performed. Histopathology confirmed lymphangioma. At present, the infant is 5 months old, showing normal lung capacity and unremarkable gastroenterology follow-up.

CONCLUSION: Prenatal diagnosis and follow-up of intraabdominal cystic lesions remain challenging. Although rare, lymphangioma should be considered in the differential diagnosis. This case underscores the importance of early detection, careful surveillance, and a multidisciplinary approach, including timely surgical management.

Keywords: fetal intraabdominal cyst, lymphangioma, neonatal intensive care, neonatal surgery, prenatal diagnosis, ultrasonography

fetal intraabdominal kistik ve kitle lezyonlar



PS-012

Fetal Cardiac Anomaly: A Case of Sotos Syndrome

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We present the case of a 29-year-old pregnant woman who was referred for further evaluation due to an increased risk for trisomy 21 identified on second-trimester screening. Genetic microarray analysis initially revealed a duplication in chromosome 1q43 encompassing the MTR and partial RYR2 genes, which was classified as a variant of uncertain significance (VUS). Prenatal ultrasound and fetal echocardiography demonstrated bilateral renal pelvis dilatation, a dysplastic pulmonary valve with mild pulmonary stenosis and regurgitation, and a small primum atrial septal defect. At 35 weeks of gestation, the fetus developed signs of cardiac failure and hydrops, necessitating an emergency cesarean delivery. Postnatal whole-exome sequencing identified a heterozygous nonsense variant in NSD1 (c.3958C>T, p.Arg1320*), classified as pathogenic and diagnostic for Sotos syndrome. This case highlights the evolving nature of genetic interpretation, the need for ongoing follow-up when VUS are identified, and the importance of integrating prenatal imaging with advanced genomic testing.

Keywords: fetal pulmonary stenosis, Sotos Syndrome, WES

PS-013

A mother diagnosed with 47 XXX and a fetus with diaphragmatic hernia diagnosed with duplication in the 4th chromosome during pregnancy: A case report

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Chromosomal anomalies such as trisomies 18, 13, and 21, microdeletion/duplication syndromes have been associated with CDH.. Here we introduce a CASE: A mother diagnosed with 47 XXX and a fetus with diaphragmatic hernia diagnosed with duplication in the 4th chromosome during pregnancy:

A Case Report A 24-year-old G1P0 patient, referred to the Maternal Fetal Medicine at 25 weeks and 6 days of gestation due to diaphragmatic hernia.The NIPT test was performed and a 31 Mb duplication of the 4q32.1–4q35.3 region was detected in NIPT..A cordocentesis procedure was performed and karyotype result was reported that 46,--der(1),t(1;4)(p36.3;q32.1)dn chromosome analysis revealed a derivative structure of chromosome 1 resulting from an unbalanced translocation between the p36.3 region of chromosome 1 and the q32.1 region of chromosome 4. Maternal chromosome was reported as 47,XXX..The microarry result reported like 1p36.33 1p36.32 heterozygous mental retardation, hypotonia phenotype and 4q32.1 4q35.2 heterozygous + intrauterine growth retardation, obesity phenotype. At the 35 weeks and 4 days, a male infant, weighting 1640 gram was delivered and the baby was underwent surgery for diaphragmatic hernia on the 5th postnatal day.The pediatric metabolic diseases department was suspected peroxisomal disease.

The baby has been hospitalized in neonatal intensive care for 100 days due to respiratory distress. conclusion

Detailed genetic testing are critical for predicting prognosis in cases of CDH. Fetal DNA is only a screening test; invasive test is required for confirmation.. Maternal genetic findings (e.g., Triple X) should also be considered in counseling.

Keywords: NIPT,diaphragmatic hernia,Triple X syndrome

PS-014

Fetal Facial Lymphangioma: Prenatal Diagnosis and Postnatal Management – A Case Report

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INTRODUCTION: Lymphangiomas are benign malformations resulting from abnormal development of the fetal lymphatic system. They are most commonly located in the cervical region, while facial involvement is rare (<10%). Prenatal diagnosis is typically established through ultrasonography and fetal MRI. Early recognition is crucial for delivery planning, neonatal intensive care and surgical management.

Case Report 37-week pregnant woman was referred to our center after an ultrasound examination revealed 61 × 45 mm mass originating from the left buccal region of the fetus. Prenatal MRI confirmed Congenital Lymphatic Malformation. Although no significant cervical extension was identified, the risk of airway compression prompted a multidisciplinary evaluation involving neonatology and pediatric surgery. Preparations for a potential EXIT procedure were made, and delivery was performed via cesarean section at 39 weeks.

At birth, no airway obstruction was noted upon examination. Postnatal MRI findings supported the diagnosis of lymphangioma. The infant was followed up, and at 3 months of age, normal feeding and respiratory findings with no increase in lesion size. Lymphangioma has considered at pediatric council. Follow-up has planned.

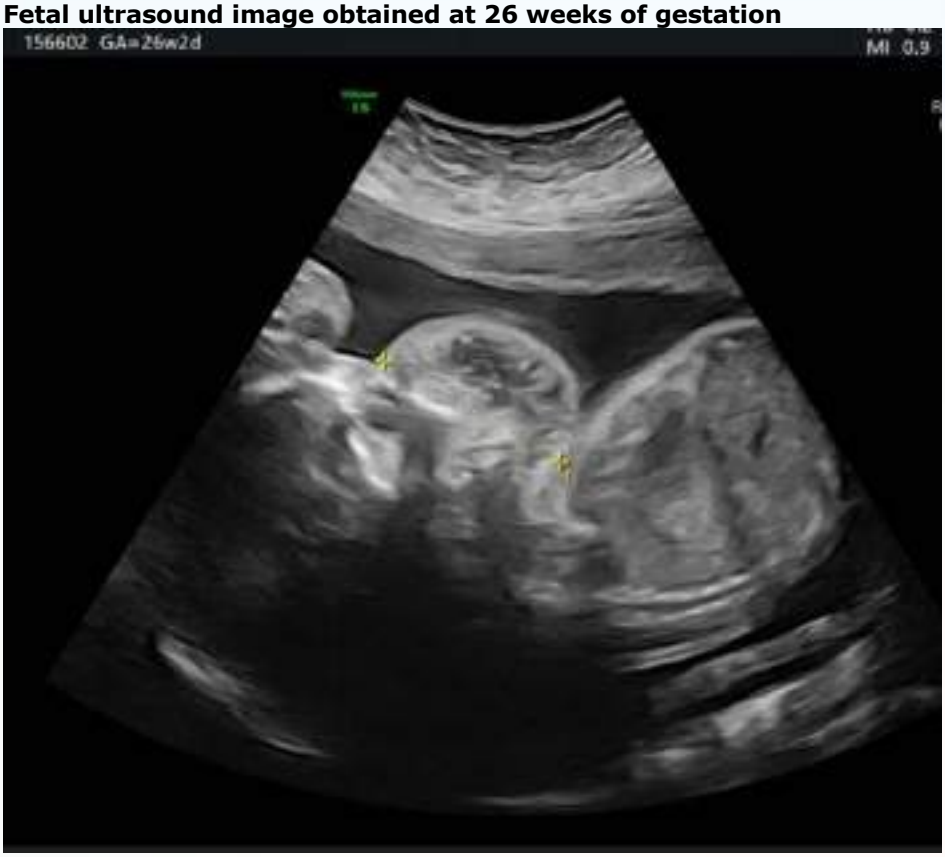
CONCLUSION: Facial lymphangiomas pose a potential risk for airway obstruction. Fetal MRI is considered the gold standard for assessing airway compromise and lesion extent. Prenatal diagnosis, multidisciplinary planning, and timely intervention lead to favorable outcomes. Airway assessment is critical in cases of facial lymphangioma.

References

1. Berchiolli R, et al. Fetal lymphangiomas: prenatal diagnosis and management. Prenat Diagn. 2023;43(4):457–464.
2. ISUOG Practice Guidelines: Role of Ultrasound in Fetal Tumor Diagnosis, 2022.

Keywords: Lymphangioma, EXIT, Congenital Lymphatic Malformation

Fetal ultrasound image obtained at 26 weeks of gestation



Fetal ultrasound image obtained at 31 weeks of gestation

Fetal ultrasound image obtained at 38 weeks of gestation

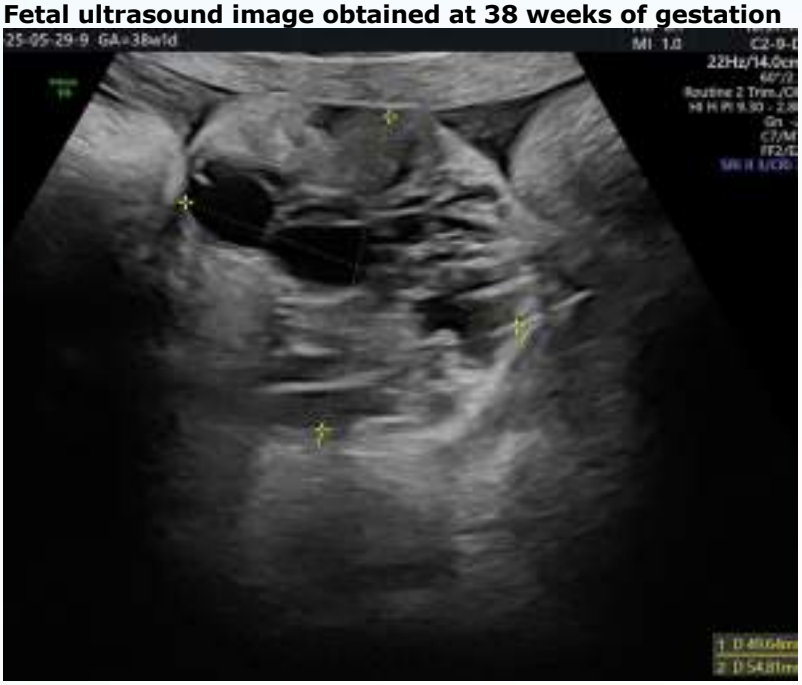


Figure 1. Postnatal clinical image showing the facial lymphangioma.

Figure 2. Postnatal clinical image showing the facial lymphangioma.

Figure 3. Postnatal MRI showing the facial lymphangioma.

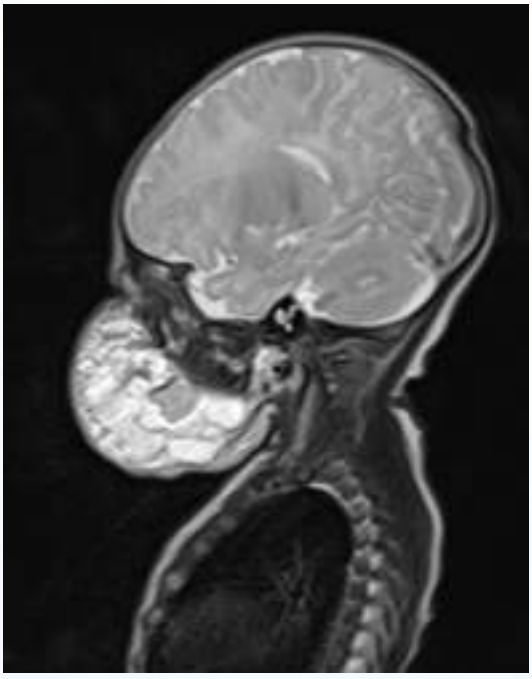
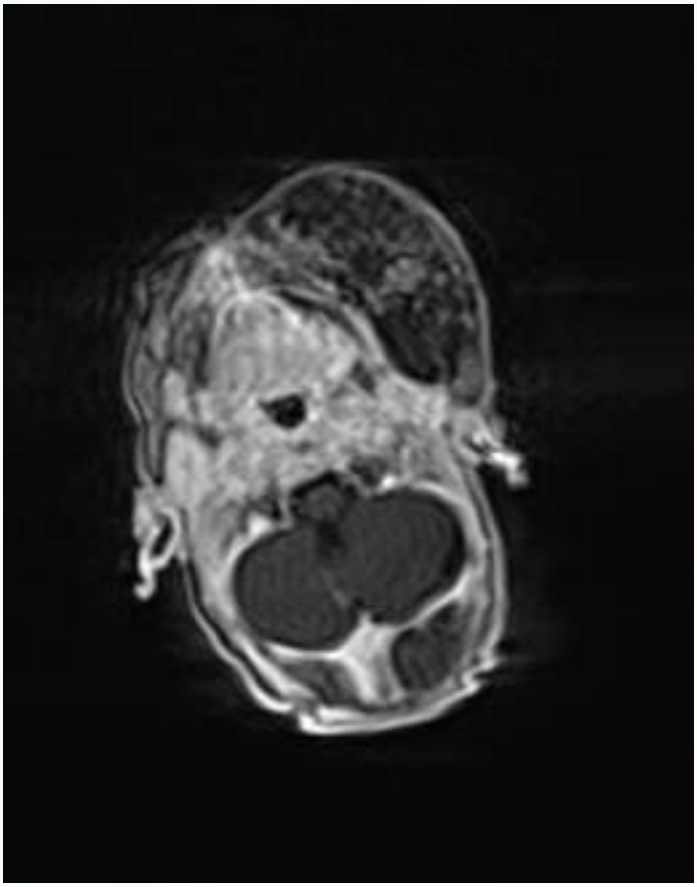


Figure 4. Postnatal MRI showing the facial lymphangioma.



PS-015

Prenatal Dönemde Fetal Pelviste Kitle Olgusu

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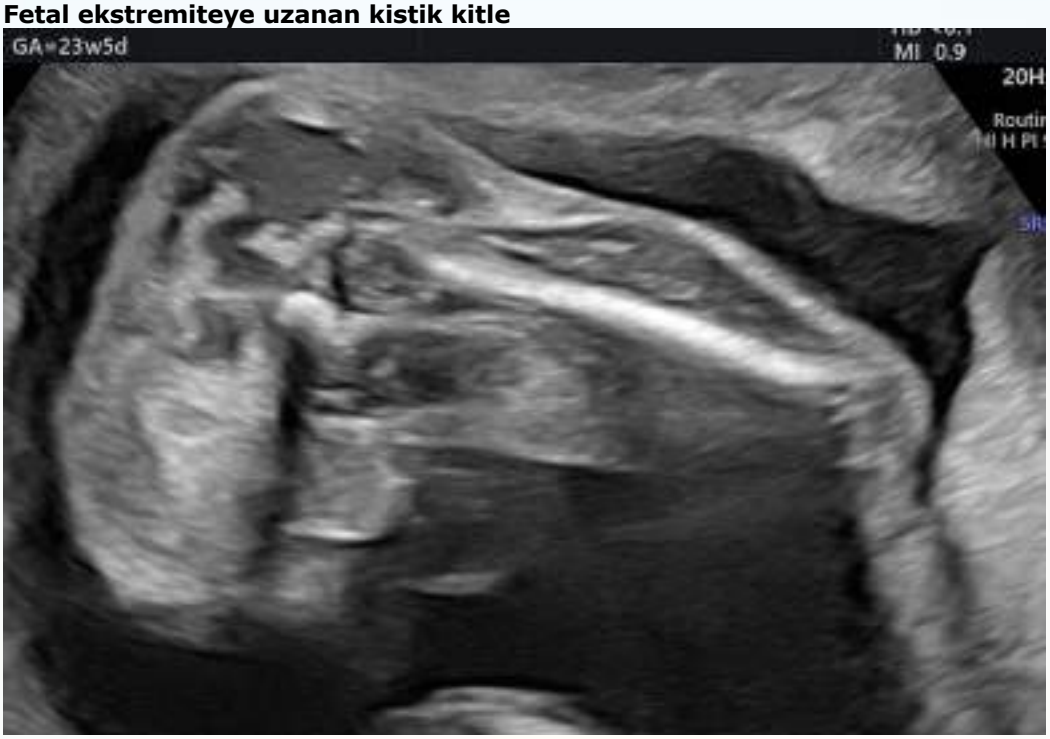
AMAÇ: Bu yazıda fetal pelviste septalı kistik kitle olgusunun sunulması amaçlanmaktadır.

BULGULAR: 24 yaşında primigravid, 23 hafta- 4 gün gebe hastanın ultrasonografi(US) muayenesinde fetal pelviste her iki gluteal bölgeye yayılan transvers çapı 50 mm, ön arka çapı 29 mm, yüksekliği 29 mm ölçülerinde içerisinde Doppler ile yoğun kan akımı izlenmeyen ön planda sakrokoksigeal teratom düşündüren kitle görüldü. Lenfovasküler malformasyon ayırıcı tanıda yer almaktaydı. Anal sfinkter görülmüş olup bu kitlenin etkisi ile mesane öne itilmiş izlendi. Ayrıca yine bu kitlenin basısına bağlı olduğu düşünülen mesane arkasında, vajina introitusundan göbek altına kadar uzanan ortadan iki bölüm halinde bölünmüş uzunluğu 40 mm, soldan sağa 25 mm, ön arka 16.5 mm ölçülerinde hidrometrokolpos olduğu düşünülen yapı izlendi. Pelvis renalis AP çapı solda 6.5 mm, sağda 3.4 mm olarak ölçüldü. Böbrek ekojeniteleri, kaliksler normal olarak değerlendirildi. Kortikal kist saptanmadı. Diğer sistemlere ait majör yapısal anomali saptanmadı. MCA PSV:29 cm/sn (0.99 MoM) ölçüldü. Genetik danışma verilerek karyotip, array, WES yapılması önerildi. MR ‘’Sol parakolik lojda en geniş yerinde 1.8x3.3 cm boyutlarında ulaşan makrolobule konturlu multiseptalı kistik natürde kitle lezyon mevcut olup ayırıcı tanıda öncelikle lenfanjiyom düşünülmüştür.’’ olarak raporlanmıştır. Postnatal dönemde yapılan **muayenesinde** Lenfovasküler malformasyon düşünülmüştür.

SONUÇ: Fetal lenfanjiomlar, nadir ve genellikle iyi huylu tümörlerdir. Ancak hızlı büyüme ve çevre dokulara invazyon potansiyeli nedeniyle prenatal tanı ve yönetim önemlidir. Prognoz kitlenin büyüklüğü ve invazyon derecesine bağlı olarak değişebilir. Fetal MR ayrıcı tanıda ve lokalizasyonda kolaylık sağlamaktadır.

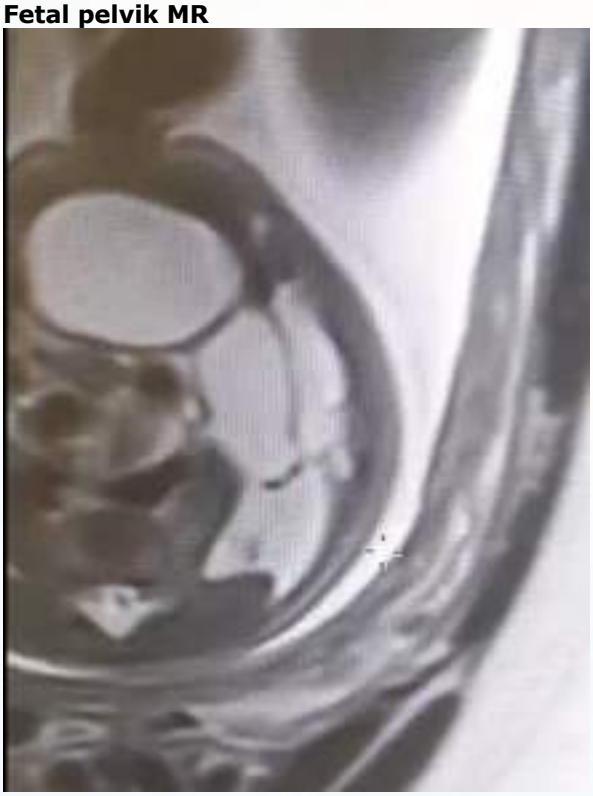
Anahtar Kelimeler: Lenfanjiom, Sakrokoksigeal teratom, fetal pelvik kitle

Fetal ekstremiteye uzanan kistik kitle



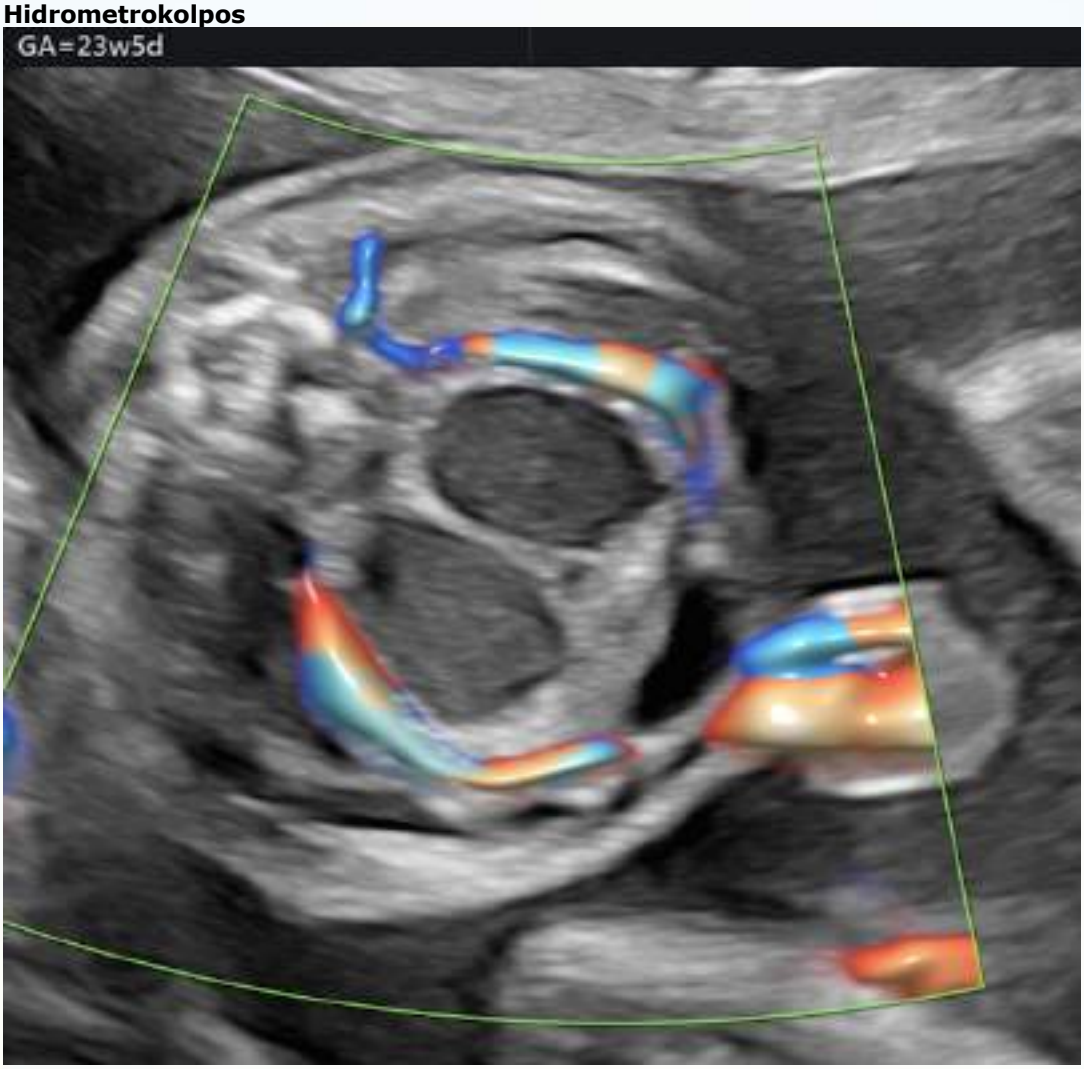
Alt ekstremiteye pelvisten uzanım gösteren belirgin çap farkına neden olmayan kistik kitle.

Fetal pelvik MR



Fetal MR'da koronal kesitlerde kistik kitlenin abdomene uzanımı ve diğer organlarla ilişkisi gözlenmektedir

Hidrometrokolpos



TAUS'de fetal pelvisin aksiyel plan kesitlerinde septasyon gösteren, her iki umblikal arterin arasında yerleşmiş kistik kitle imajı ön planda female fetüs olması sebebi ile hidrometrokolpos olabileceği düşünülmüştür.

PS-016

Prenatal tanılı Dakriyosistoselde spontan rezolüsyon

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Trakya Üniversitesi, Trakya Tıp Fakültesi, Kadın Hastalıkları ve Doğum, Perinatolojı ABD, Edirne

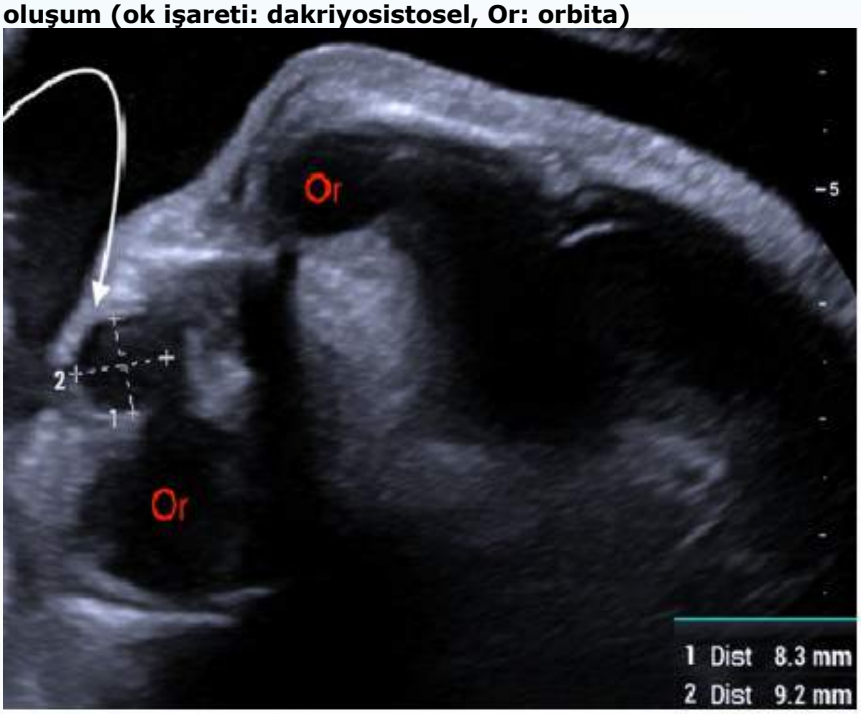
GİRİŞ: Doğumsal dakriyosistosel, nazolakrimal kanalın distal kısmındaki tıkanıklığa bağlı olarak orbita medialinde kistik kitle şeklinde ortaya çıkan, nadir bir durumdur. Prenatal dönemde tipik olarak tek taraflı, iyi sınırlı, ince duvarlı ve anekoik kistik lezyon şeklinde izlenir. Antenatal dönemde saptanan dakriyosistoselli çoğu olguda doğum sonrası lezyon spontan olarak gerilemektedir. Nadiren solunum sıkıntısı veya enfeksiyon gibi neonatal komplikasyonlarla birlikte bulunabilir. Prenatal tanı, doğum sonrası neonatal takip açısından önemlidir.

OLGU: 37 yaşında G2P0A1 olan gebenin 21. haftada yapılan detaylı USG değerlendirmesinde herhangi bir özellik görülmedi. Gebenin 30 hafta 1 günlük iken yapılan usg değerlendirilmesinde fetusta sağ orbita medialinde 9,2 × 8,3 mm boyutlarında anekoik, ince duvarlı kistik yapı tespit edildi ve dakriyosistosel olarak değerlendirildi (Figür 1 ve 2). Olgu perinatoloji konseyinde görüşüldü ve antenatal dönemde başka ek anomali veya komplikasyon gelişmedi. Gebelik 38. haftaya kadar düzenli aralıklarla izlendi ve bu haftada dış merkezde 3200gr sağlıklı tek canlı kız bebek sezaryen ile doğurtuldu. Yenidoğanın postnatal göz muayenesinde lezyonun spontan rezolüsyonu gözlendi.

TARTIŞMA: Prenatal dakriyosistosel nadir görülse de yüksek çözünürlüklü ultrasonografi ve özellikle üç boyutlu görüntüleme tekniklerinin yaygınlaşması ile tanı sıklığı artmaktadır. Lezyon genellikle benign seyirli olup çoğu olguda doğum sonrası ilk haftalarda kendiliğinden kaybolur. Ancak kitle nazal kaviteye uzanarak doğum sonrası nazal obstrüksiyon, solunum sıkıntısı veya dakriyosistit gibi komplikasyonlara yol açabilir. Literatürde, antenatal tanının aile bilgilendirmesi ve doğum sonrası yönetimin planlanması açısından önemli olduğu, özellikle bilateral lezyon varlığında neonatal solunum yolu tıkanıklığına karşı hazırlıklı olunması gerektiği vurgulanmaktadır. Bu olgu ile tek taraflı ve izole dakriyosistoselde lezyonun postnatal rezolüsyonunun muhtemel ola bileceği gösterilmiştir.

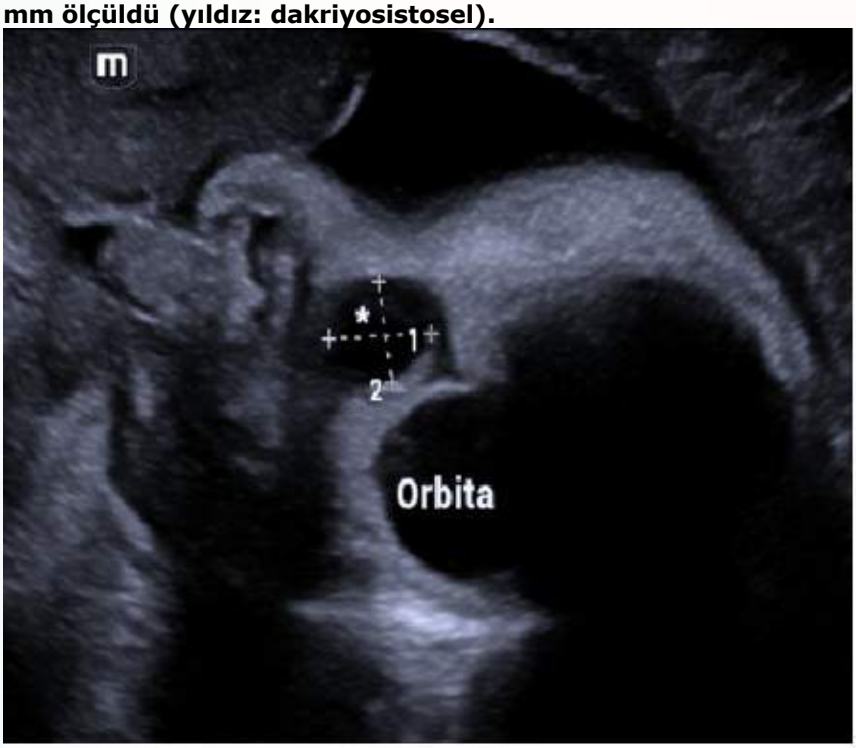
Anahtar Kelimeler: Dakriyosistosel,Göz yaşı kanalı anomalisi, Rezolüsyona uğrayan kist

Figür 1. 30 +1 hafta gebelikte sağ orbita medialinde dakriyosistosel ile uyumlu kistik oluşum (ok işareti: dakriyosistosel, Or: orbita)



30 +1 hafta gebelikte sağ orbita medialinde dakriyosistosel ile uyumlu kistik oluşum (ok işareti: dakriyosistosel, Or: orbita)

Figür 2: 34. gebelik haftasında parasagital planda dakriyosistosel ile uyumlu lezyon 9,6 x8,4 mm ölçüldü (yıldız: dakriyosistosel).



34. gebelik haftasında parasagital planda dakriyosistosel ile uyumlu lezyon 9,6 x8,4 mm ölçüldü (yıldız: dakriyosistosel).

PS-018

Dichorionic Diamniotic Twin Pregnancy, Neural Tube Defect In One Fetus: A Case Report

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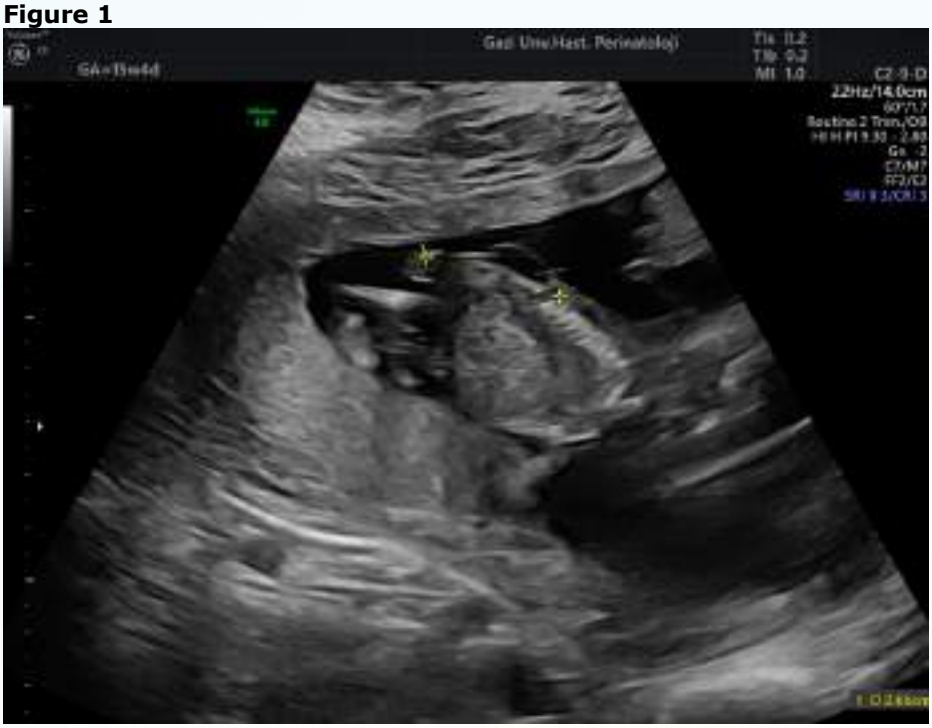
If multiple fetuses are discordant for anatomical or genetic anomalies,elimination of the abnormal fetüs is an option.Selective termination (ST) refers to a procedure in which one or more specific fetuses of a multifetal gestation are terminated due to a known or suspected fetal abnormality,including a chromosomal,structural,genetic or growth abnormality.

In this case report,we present a 34-year-old,gravida 2 para 1 pregnant patient with neural tube defect in one fetus. At 14 weeks and 2 days of gestation, ultrasonographic examination revealed flexion contractures of the lower extremities in the upper fetus. Prenatal diagnostic testing was offered,but the patient declined.At 15 weeks and 2 days of gestation,ultrasonography revealed a lumbosacral neural tube defect, sacral agenesis,lower extremity flexion contractures and pes equinovarus in the upper fetus.With informed constent of the family,at 16 weeks and 3 days of gestation selective termination was performed for the fetus with anomalies. Amniocentesis was performed for the lower fetus following a high biochemical risk for trisomy 21 in first-trimester screening;results were normal.The patient, followed regularly in our clinic,is currently at 29 weeks of gestation with no reported complications. Because abnormalities are often not fully delineated until the second trimester,selective termination(ST) is performed later in gestation than selective reduction and entails greater risk.This procedure is therefore usually not performed unless the abnormality is severe but not lethal.In continuing pregnancies,outcome after ST is generally favorable,with low rates of prematurity and few maternal complications.

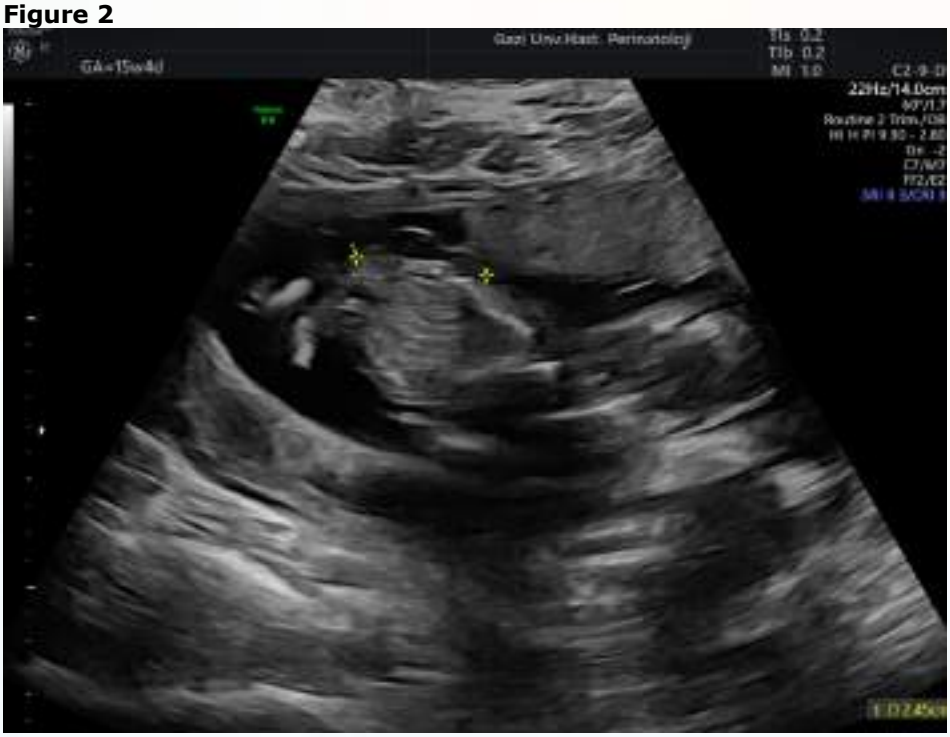
Keywords: Neural tube defect,selective termination,twin pregnancy

Figure 1



Neural tube defect

Figure 2



Neural tube defect

PS-019

Gebelikte prenatal tanısı konulan mekonyum peritoniti: nadir bir olgu sunumu

Zeliha Kurtoğlu Kuzucu[1], Şule Birol İnce[1], Filiz Yarşılıkal Güleroğlu[2]
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GİRİŞ: Mekonyum peritoniti, intrauterin bağırsak perforasyonu sonucu mekonyumun periton boşluğuna geçmesiyle gelişen nadir fakat ciddi bir tablodur. Prevalansı yaklaşık 1/30.000 doğum olarak bildirilmiştir. Prenatal tanı, doğumun yönetimi ve neonatal bakım açısından kritik öneme sahiptir. Bu yazıda prenatal dönemde tanısı konulan ve başarıyla yönetilen bir olgu sunulmaktadır.

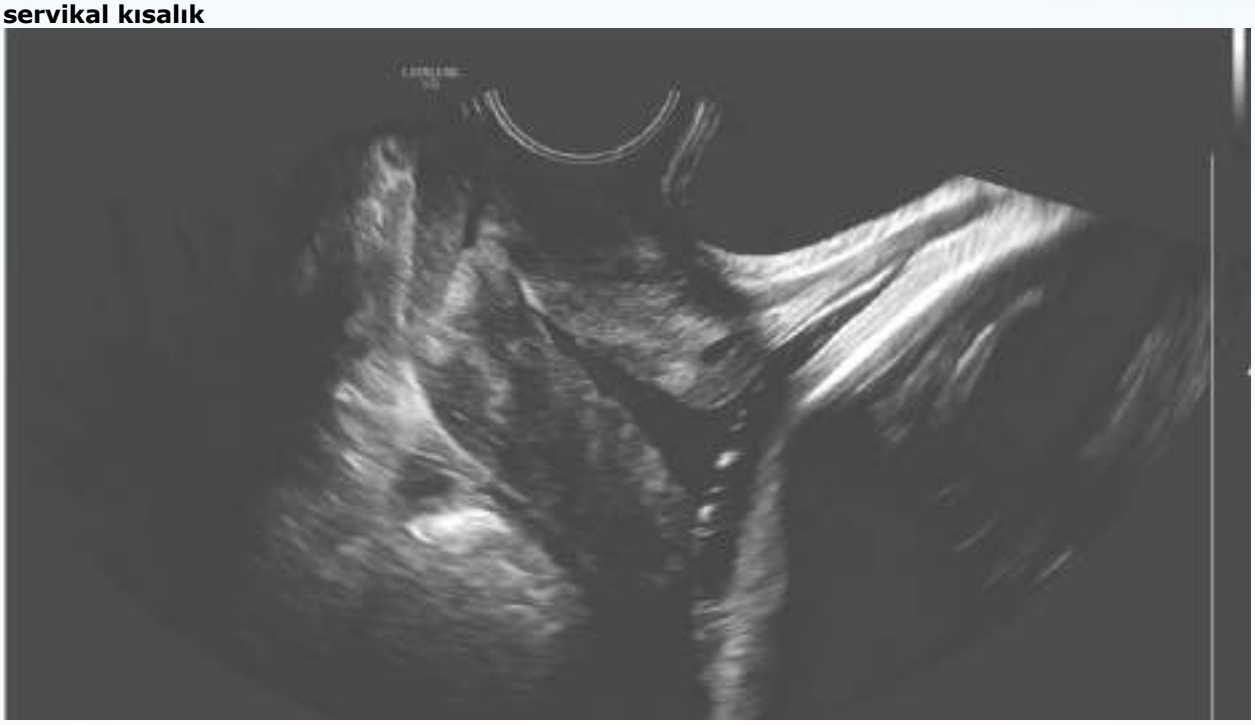
OLGU: 26 yaşında, gravida 1, parite 0 olan hasta, 33+2 gebelik haftasında dış merkezden mekonyum peritoniti şüphesiyle tarafımıza yönlendirildi. Ultrasonografide batın içinde yaygın kalsifikasyonlar, polihidramnios ve hidrosel izlendi; belirgin bağırsak dilatasyonu saptanmadı. Target sign görüldü. NST'de kontraksiyon ve servikal kısalma olması üzerine hasta erken doğum tehdidi tanısıyla yatırıldı. Tokolitik tedavi ve betametazon uygulandı. 33+6 haftada servikal açıklığın ilerlemesi nedeniyle vajinal doğum gerçekleşti. Yenidoğan mekonyum peritoniti ön tanısıyla yoğun bakım ünitesinde izlendi. Abdominal grafi ve bilgisayarlı tomografide yaygın intraabdominal kalsifikasyonlar görüldü. Pediatrik cerrahi tarafından değerlendirilen olguda cerrahi girişim gereksinimi olmadığına karar verildi. Medikal tedavisi tamamlanan bebek sağlıklı şekilde taburcu edildi.

TARTIŞMA: Prenatal mekonyum peritoniti tanısı nadirdir. Ultrason bulguları, özellikle intraabdominal kalsifikasyonlar ve polihidramnios, tanıda yol göstericidir. Erken tanı multidisipliner planlama olanağı sağlar ve tüm olgular cerrahi girişim gerektirmez.

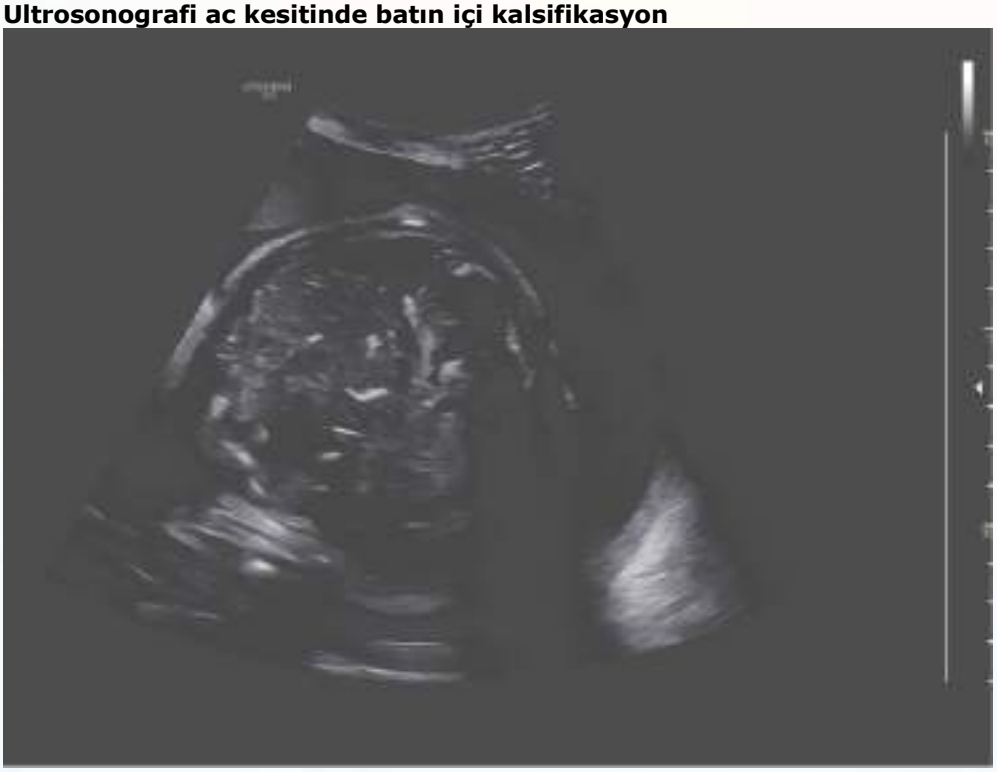
SONUÇ: Prenatal mekonyum peritoniti tanısı, perinatal yönetim ve neonatal sonuçlar açısından önemlidir. Bu olgu, erken tanı ve multidisipliner yaklaşımın değerini vurgulamaktadır

Anahtar Kelimeler: Mekonyum peritoniti, Prenatal tanı, Fetal ultrasonografi

servikal kısalık



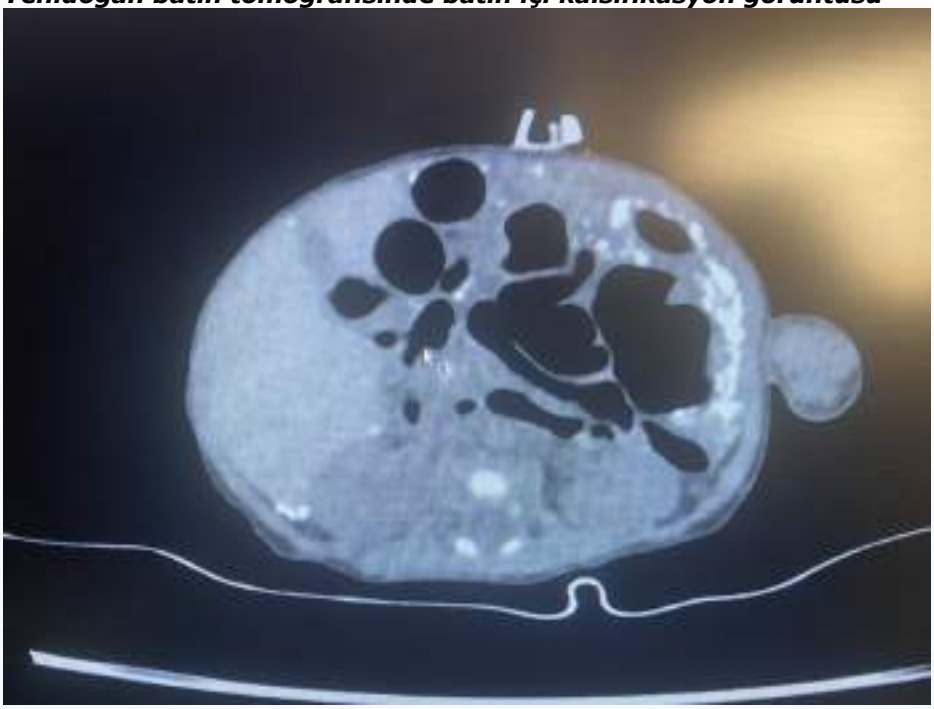
Ultrosonografi ac kesitinde batın içi kalsifikasyon



Yenidoğaan abdominal grafide batın içi kalsifikasyonlar

Yenidoğana ait direkt batın grafisinde, karın grafsinde yaygın ve belirgin radyoopak alanlar izlenmektedir Bu bulgular, intrauterin bağırsak perforasyonu sonrasında periton boşluğuna sızan mekonyumun kalsifikasyonları ile uyumludur.

Yenidoğan batın tomografisinde batın içi kalsifikasyon görüntüsü



PS-021

Prenatal Diagnosis of Isolated Cleft Lip and Palate: A Case Report with Postnatal Surgical Planning and Multidisciplinary Follow-up

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BACKGROUND: Cleft lip and/or palate (CL/P) is among the most frequent congenital craniofacial anomalies, with an estimated prevalence of 1–2 per 1,000 live births. Prenatal diagnosis allows for early family counseling, genetic assessment, and multidisciplinary planning. We report a prenatally diagnosed case of isolated cleft lip and palate with comprehensive antenatal and postnatal follow-up.

CASE: A 26-year-old primigravida presented for routine antenatal care. At 21+1 weeks' gestation, detailed ultrasound revealed unilateral cleft lip with probable palate involvement. Fetal neurosonography and echocardiography excluded additional malformations. Invasive testing was performed: QF-PCR and chromosomal microarray were normal; TORCH screening was negative. The pregnancy proceeded uneventfully, with appropriate fetal growth and reassuring biophysical parameters. At term, a male infant weighing 2760 g was delivered via cesarean section. Postnatal evaluation confirmed a complete unilateral cleft lip and palate. No additional anomalies were detected, and genetic analysis remained normal. The neonate was admitted to the NICU for feeding support and multidisciplinary assessment including neonatology, plastic surgery, ENT, and genetics. Surgical repair planning was initiated during early infancy.

CONCLUSION: This case emphasizes the significance of systematic fetal anatomical scanning in mid-gestation for early detection of craniofacial anomalies. Accurate prenatal diagnosis of isolated CL/P improves parental preparedness, enables genetic evaluation, and optimizes perinatal and postnatal care. Multidisciplinary follow-up remains essential for surgical, nutritional, and developmental outcomes.

Keywords: cleft lip, cleft palate, prenatal diagnosis, fetal anomaly, congenital malformation, antenatal care

Postnatal diagnosis of cleft lip/palate

At term, a male infant weighing 2760 g was delivered via cesarean section. Postnatal evaluation confirmed a complete unilateral cleft lip and palate.

Prenatal diagnosis of cleft lip/palate with USG

At 21+1 weeks' gestation, detailed ultrasound revealed unilateral cleft lip with probable palate involvement.

PS-022

TMEM67 Homozigot Mutasyonu ile ailesel tekrarlayan Joubert sendromu: prenatal tanı ve yaklaşım

Türkan Kızgın, Rashad Mammadov, Emre Göktürk Çelik, Cihan İnan, Cenk Sayın
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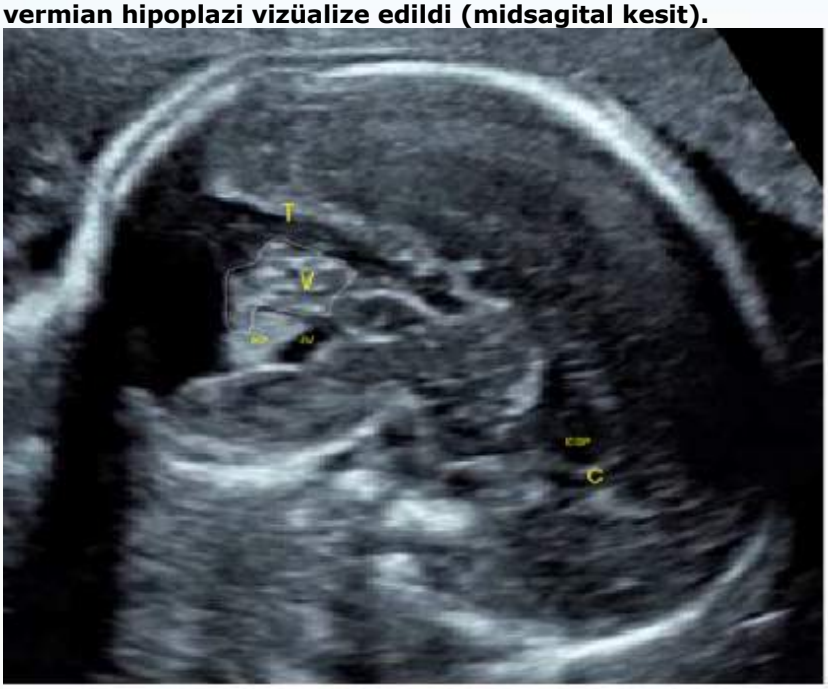
Joubert sendromu (JS),serebellar vermis hipoplazisi/dizgenezisi, beyin sapında "molar tooth" işareti ve değişken sistemik bulgularla karakterize OR geçişli nadir bir siliopatidir.Prenatal tanıda posterior fossa genişliği, vermian hipoplazi ve serebellar ayrışma en önemli bulgulardır.Güncel kılavuzlar, şüpheli olgularda hedefli nörosonografi ve fetal MRG'nin birlikte kullanılmasını; ek anomaliler varlığında karyotip,CNV veya hedef gen paneli/WES temelli testleri önermektedir.TMEM67 mutasyonları Joubert–Meckel spektrumunda renal/hepatik tutulum riskini artıran alt grup olarak tanımlanmıştır. Vakamız aynı mutasyonun iki kardeşte tekrarlandığı olguyu sunmaktadır.

32 yaşında,G2P1,2.derece kuzen evliliği ve ilk çocuğunda JS tanısı bulunduğu için gebeliğinin 21. haftasında ayrıntılı ultrasonografi için kliniğimize başvurdu. USG'de bilateral koroid pleksus kistleri, arka fossada belirgin dilatasyon (CM:11mm) ve serebellar vermis hipoplazisi,serebellar ayrışma saptandı.Bilateral böbrekte hiperekojenite ile kortikomedüller ayrım kaybı izlendi.22.haftada perinatoloji konsey invaziv prenatal tanı(amniyosentez) ve fetal MRG önerdi.Amniyosentez sonucu,TMEM67 (NM_153704.6) c.413G>T (p.Arg138Ile)mutasyonunun homozigot olduğu gösterildi.Anne ve baba heterozigot taşıyıcıydı ve bu mutasyonun ailede Joubert sendromu tanılı 12 yaşındaki çocukta da aynı şekilde bulunduğu biliniyordu.26. gebelik haftasında USG'de posterior fossanın genişlemeye devam ettiği(CM:12,5 mm)serebellar vermisin belirgin hipoplazik olduğu izlendi.Lateral ventriküller arasında asimetri gözlendi(sol 3,7 mm; sağ 7,3 mm).Fetal MRG'de sulkal gelişimin 22 haftalık gebelikle uyumlu olduğu,korpus kallozumun splenium bölümünde hipoplazi bulunduğu ve arka bölümün net seçilemediği saptandı.Mega sisterna magna izlendi (sisterna magna 12,5 mm).Önceki gebelikteki Joubert sendromlu çocukta saptanan aynı TMEM67 homozigot mutasyon doğrulandı.Perinatoloji konseyinde gebelik terminasyonu önerildi; aile gebeliğin devamını isteyerek izleme alındı.

Posterior fossa anomalilerinde nörosonografi ve MRG'nin tanısal değeri yüksektir.TMEM67 mutasyonu iki kardeşte nüks ederek otozomal resesif kalıtımı ve %25 tekrar riskini doğrulamaktadır.Olgumuz, öykü ile hedefe yönelik genetik doğrulamanın gebelik yönetiminde kritik rolünü vurgulamaktadır.

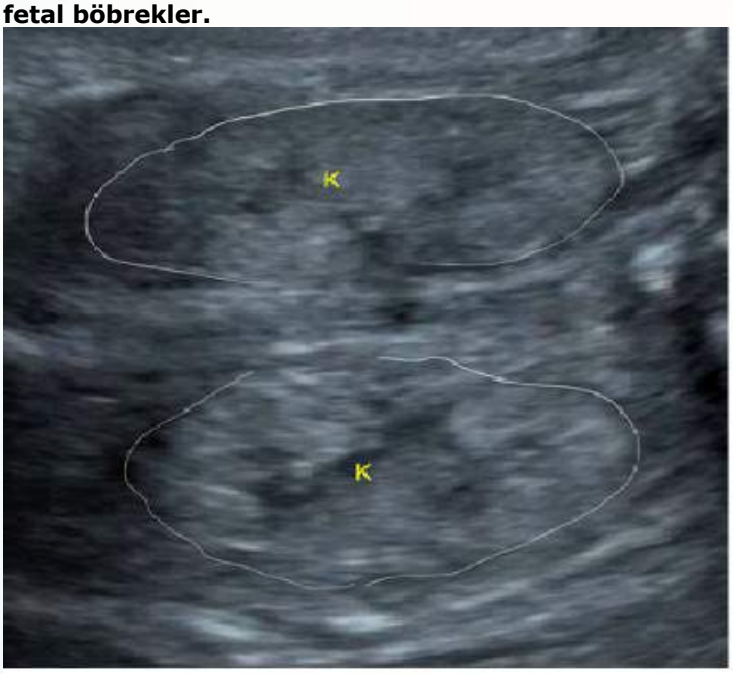
Anahtar Kelimeler: Joubert sendromu, Mega Sisterna Magna, TMEM67 gen mutasyonu

Figür 1: 21+6 hafta gebede yapıaln ultrasonografide fetal arka fossa genişlemiş izlendi ve vermian hipoplazi vizüalize edildi (midsagital kesit).



21+6 hafta gebede yapıaln ultrasonografide fetal arka fossa genişlemiş izlendi ve vermian hipoplazi vizüalize edildi (midsagital kesit). C: korpus kallozum, CSP: kavum septi pellisidum, V: vermis, 4Vve KP: 4. ventrikül ve koroid pleksusu, T: tentorium cerebelli.

Figür 2: İlk çocuğunda Joubert sendromu tanısı mevcut, 21+6 hafta gebede bilateral fetal böbrekler büyük, hiperekojen izlendi, ek olarak kortikomedüler ayırımı net yapılamadı. K: fetal böbrekler.



İlk çocuğunda Joubert sendromu tanısı mevcut, 21+6 hafta gebede bilateral fetal böbrekler büyük, hiperekojen izlendi, ek olarak kortikomedüler ayırımı net yapılamadı. K: fetal böbrekler.

PS-023

Prenatal Diagnosis and Management of Open Spina Bifida: A Case-Centered Review

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Open spina bifida remains a disabling neural tube defect despite advances in prenatal screening. We present a second-trimester fetus with typical cranial markers—the lemon sign and a crescent-shaped cerebellum—together with a lumbosacral defect on targeted neurosonography. Amniocentesis was performed; chromosomal microarray results were pending at counseling.

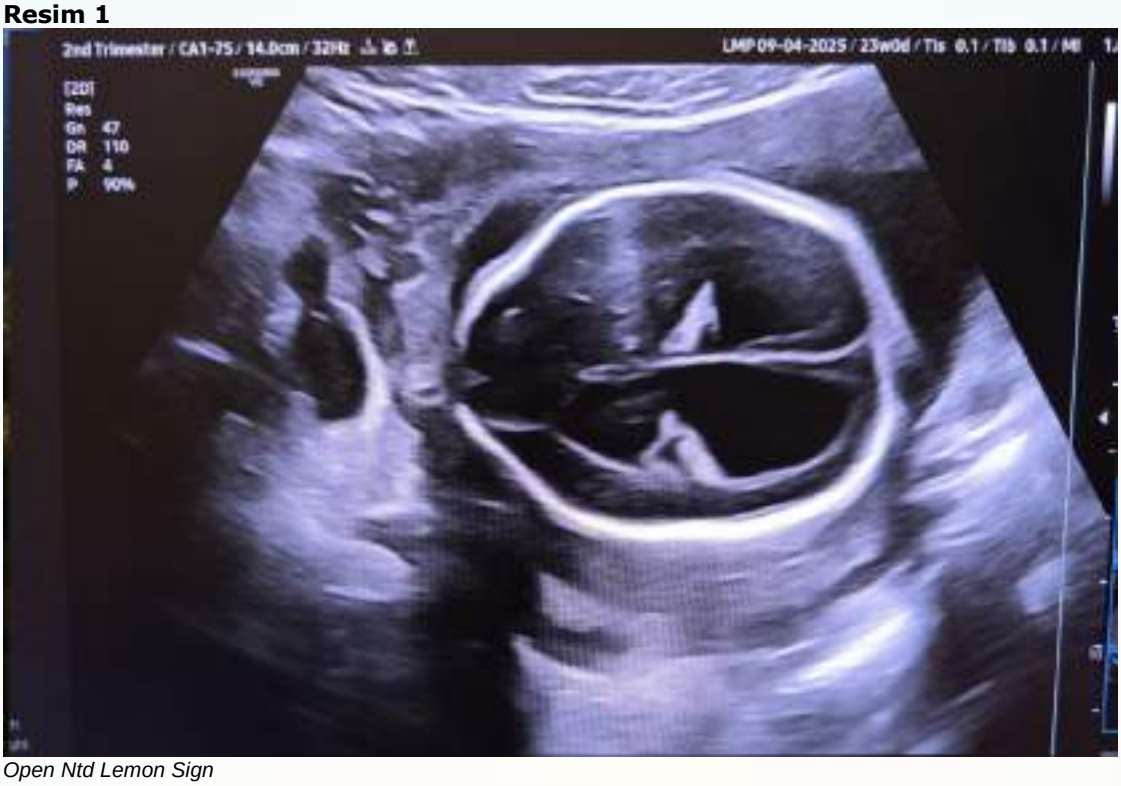
Management centered on three pathways. Prenatal closure in a specialized center was discussed, offering fewer ventriculoperitoneal shunt placements and improved motor outcomes, balanced against maternal laparotomy, hysterotomy, preterm delivery, and mandatory cesareans. Postnatal repair was considered widely accessible, though long-term mobility and continence depend on lesion level, ventricular size, and associated anomalies. Pregnancy termination, where legally permitted, was also addressed in cases with poor prognosis or limited access to fetal therapy.

Counseling emphasized key issues parents raise: likely gestational age at delivery under each option, neonatal course in the first 72 hours, and long-term needs such as catheterization, orthotics, and shunt surveillance. Pending genetic results were noted for their potential to alter prognosis if a syndromic diagnosis emerged.

This case illustrates that optimal management is not uniform but shaped by evidence, resources, and family values. Multidisciplinary, individualized counseling remains the cornerstone of care.

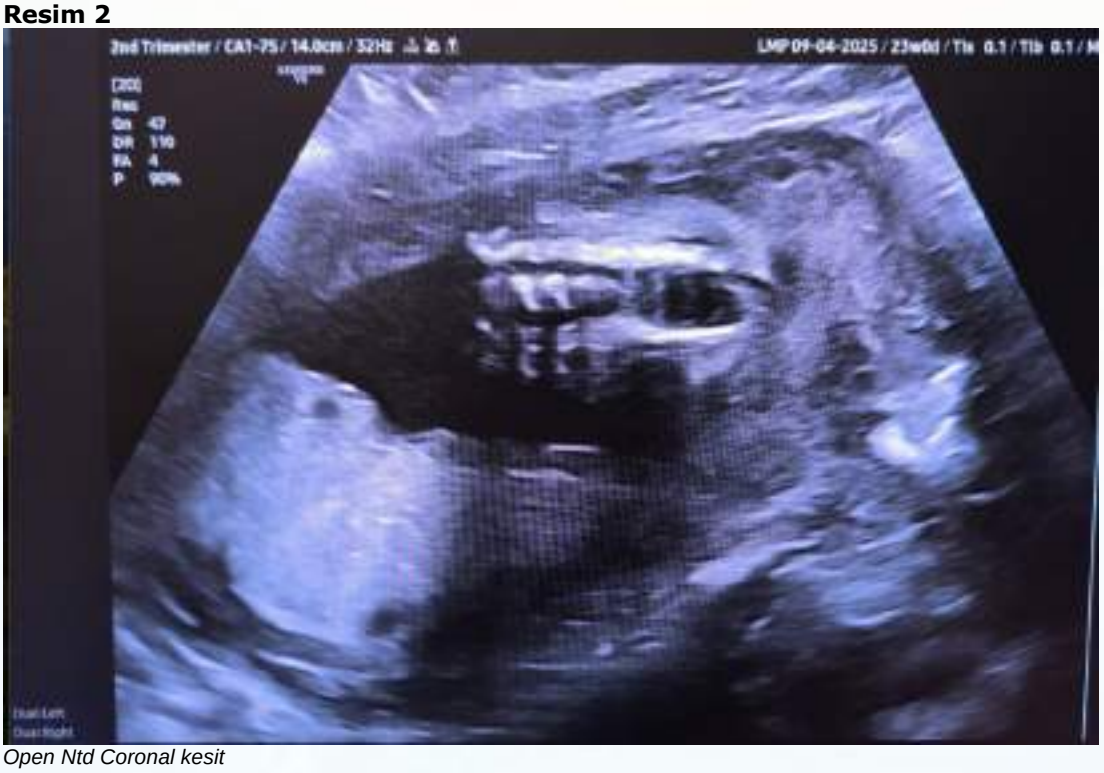
Anahtar Kelimeler: doğum öncesi tanı, fetal cerrahi, spina bifida

Resim 1



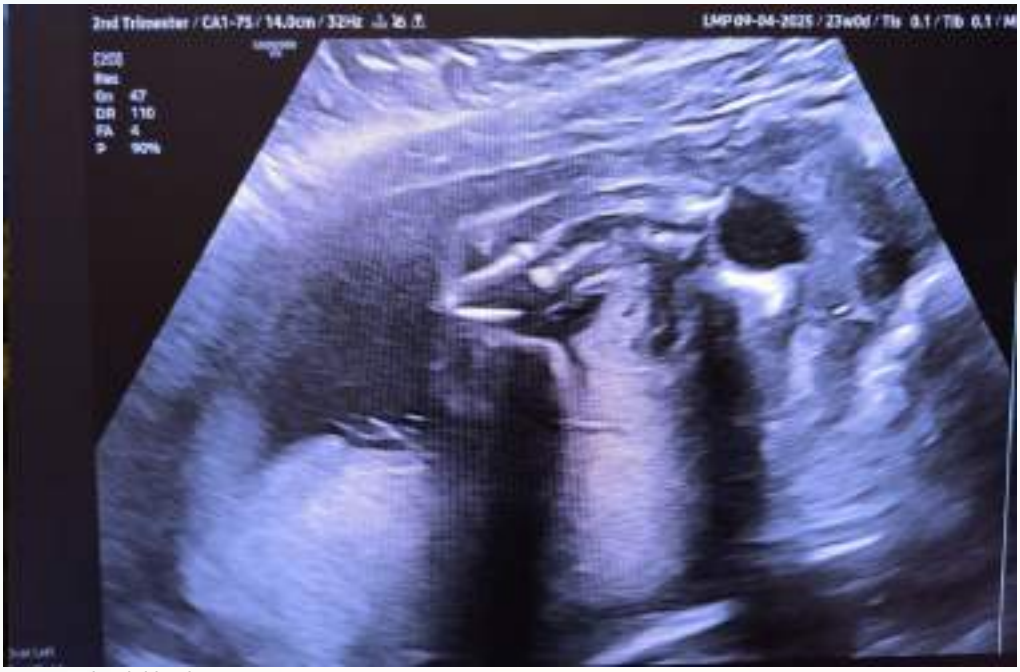
Open Ntd Lemon Sign

Resim 2



Open Ntd Coronal kesit

Resim 3



Open Ntd Axial kesit

Resim 4

Torakolumbar açık-geniş Ntd görüntüsü

Resim 5

Torakolumbar açık-geniş Ntd

PS-024

Fetal Congenital Heart Defect, Double-Outlet Right Ventricle (DORV): A Case Report

Hatice Aydın Salman, Arzu Sümer Ortaç, Esra Nuhoğlu Bilgin, Esin Şahin Toruk, Bilal Emre Erzeneoğlu, Pınar Tokdemir Çalış, Deniz Karçaaltıncaba, Mehmet Zeki Taner, Tuncay Nas
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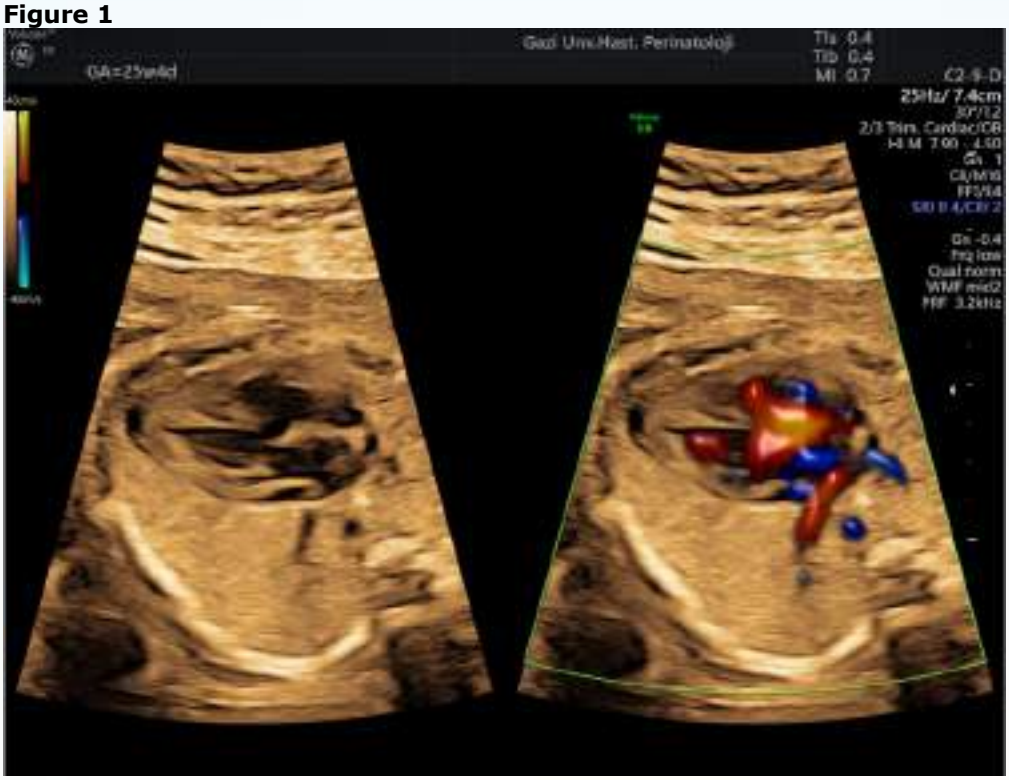
DORV is a type of ventriculoarterial connection in which both the great vessels arise either entirely or predominantly from the right ventricle.The five-chamber view is abnormal in DORV because it shows the VSD and the origin of both the great arteries from the right ventricle. Pulmonary stenosis is the most common associated malformation and occurs in about 70% of cases. Chromosomal abnormalities are frequently found in the range of 12% to 40% and primarily include trisomy 18,trisomy 13,and deletion 22q11.

In this case report,we present a 24-year-old,gravida 2 abortus 1 pregnant patient with DORV. Second-trimester screening at an outside center revealed a high risk for trisomy 21,and the patient was referred to our clinic at 23 weeks+5 days of gestation. Amniocentesis was recommended to the patient, but she refused the procedure.Antenatal fetal echocardiography revealed DORV,pulmonary artery hypoplasia, persistent left superior vena cava(LSVC),subaortik ventricular septal defect (VSD),secundum atrial septal defect(ASD),mild pulmoner stenosis. A female infant (3060 g, Apgar 8/9) was delivered vaginally at 40 weeks and 3 days of gestation.Postnatal echocardiography confirmed antenatal echocardiography findings.Postnatal chromosomal and 22q11 deletion FISH analysis were performed and found to be normal.Array analysis is ongoing.The infant is currently being followed by pediatric cardiology and cardiovascular surgery.Following angiography,corrective surgical intervention was planned for the infant.

Outcome of DORV strongly depends on the presence and severity of additional anomalies of the fetus. Surgical intervention in DORV can lead to a favorable outcome in simple DORV.The overall prognosis for fetuses with DORV is generally poor,when associated with chromosomal and extracardiac abnormalities.

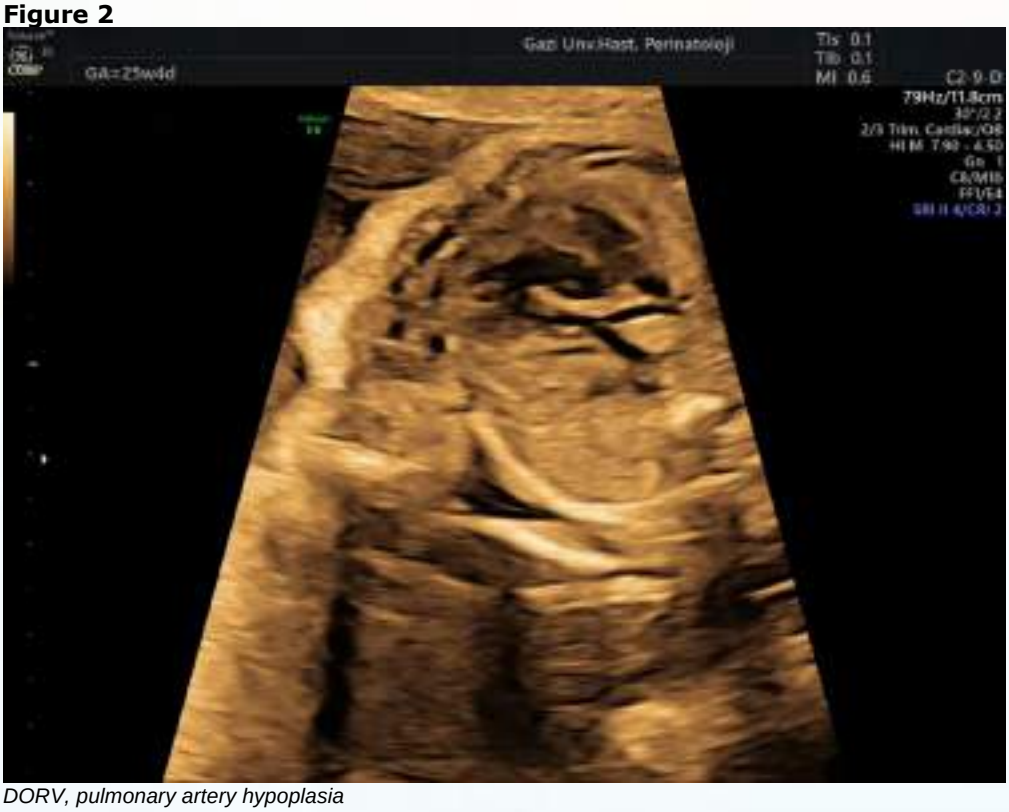
Keywords: DORV, VSD, pulmonary artery hypoplasia

Figure 1



VSD

Figure 2



DORV, pulmonary artery hypoplasia

PS-025

15 Haftalık Gebelikte Prenatal Tanı Alan Sol Kalp Hipoplazisi: Olgu Sunumu

Nigar Mammadova, Oya Demirci
Özel Klinik

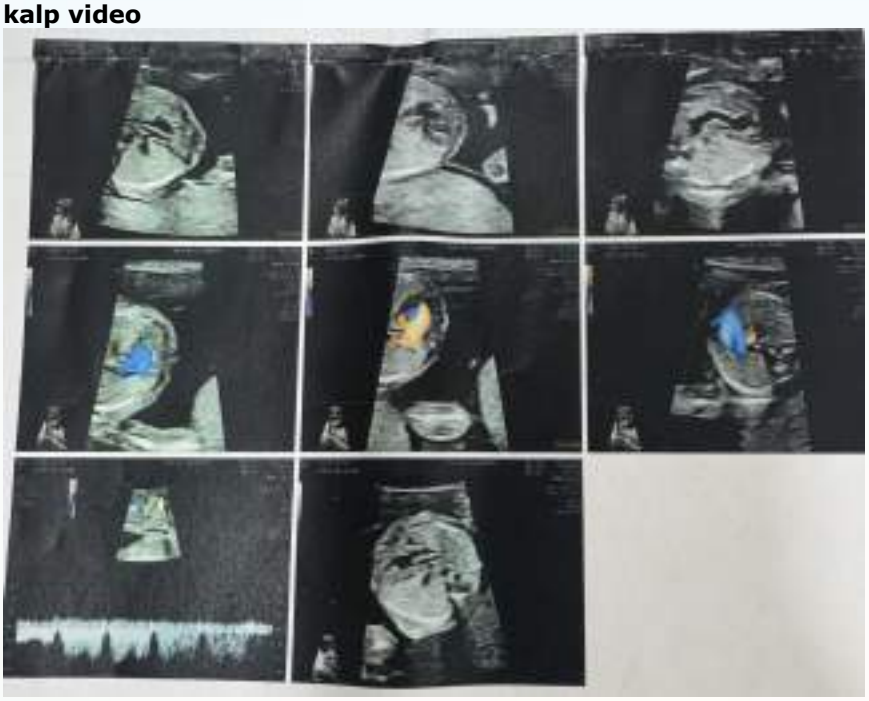
AMAÇ: Sol kalp hipoplazisi (HLHS), sol ventrikül ve sol kalp yapılarının gelişim geriliği ile karakterize, ciddi morbidite ve mortaliteye yol açan nadir bir konjenital kalp hastalığıdır. Bu olguda, 15. gebelik haftasında prenatal tanı alan nadir bir HLHS vakası sunulmaktadır.

OLGU: 26 yaşında, G3P2Y1EX1, sağlıklı kadın, 15. gebelik haftasında rutin tarama ultrasonografisinde dört boşluk görüntüsünde sol ventrikülün belirgin küçük olduğu saptandı. Fetal ekokardiyografi ile sol ventrikül hipoplazisi, mitral kapak atrezi ve aort kapak hipoplazisi doğrulandı. Ekstrakardiyak anomali saptanmadı. Aileye tanı, prognoz ve olası cerrahi seçenekler hakkında bilgi verildi. Gebelik sonlandırıldı.

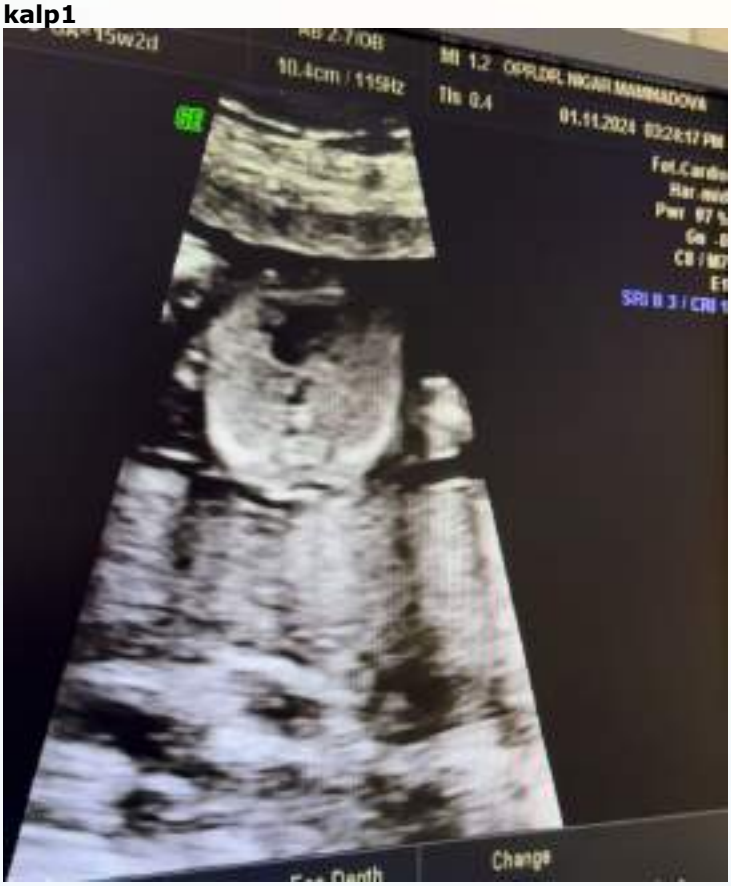
SONUÇ: HLHS'nin erken gebelik haftalarında tanınması, multidisipliner yaklaşım, doğumun uygun merkezde planlanması ve aileye danışmanlık açısından kritik öneme sahiptir. Bu olgu, 15. haftada tanı konulan nadir HLHS vakalarından biri olup, erken prenatal tanının klinik değerini vurgulamaktadır.

Anahtar Kelimeler: Fetal Ekokardiyografi, Gebelik Haftası, Konjenital Kalp Hastalıkları, Prenatal Tanı, Sol Kalp Hipoplazisi

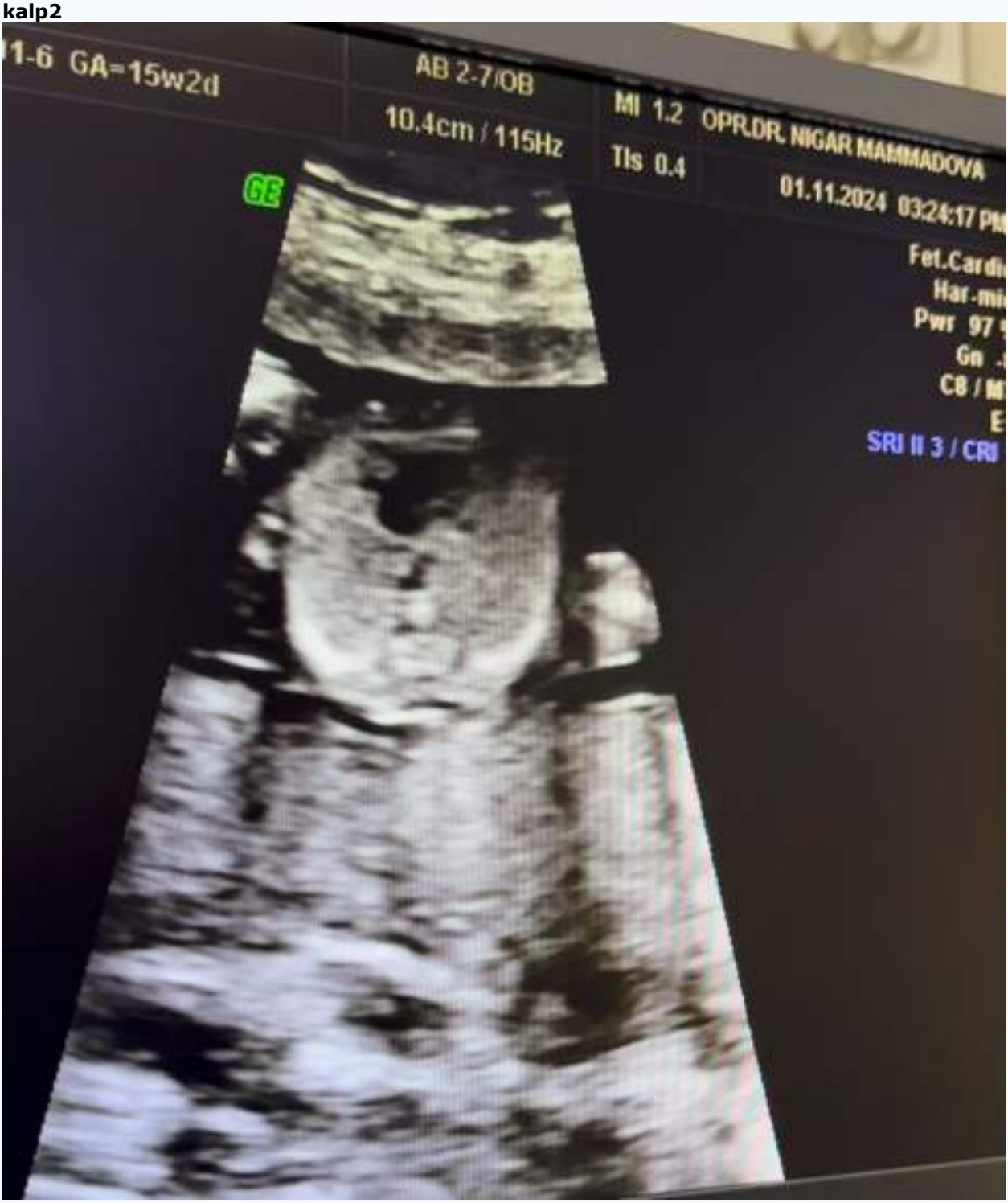
kalp video



kalp1



kalp2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PS-026

Ürogenital, Renal ve Anorektal Anomalilerin Birlikteliği Vakası

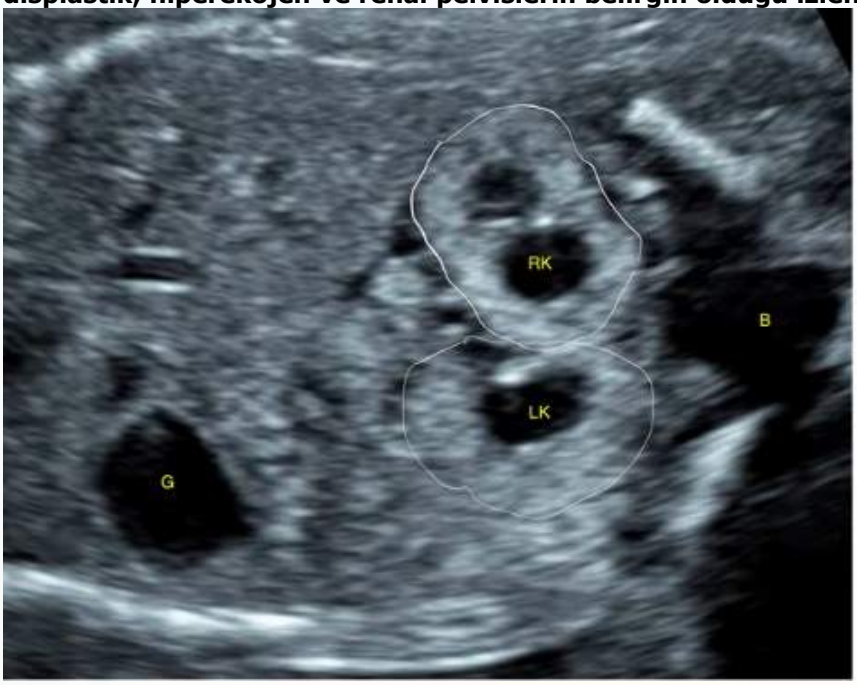
Türkan Kızgın, Rashad Mammadov, Emre Göktürk Çelik, Cenk Sayın, Cihan İnan
Trakya Tıp Fakültesi, Kadın Hastalıkları ve Doğum, Perinatolojı ABD, Edirne

Konjenital üriner sistem anomalileri sıklıkla gastrointestinal,kardiyak ve genital malformasyonlarla birlikte görülür.Prenatal dönemde erken tanı,hem prognozun öngörülmesi hem de doğum sonrası yönetimin planlanması için kritik öneme sahiptir.Ayrıntılı ultrasonografi, genetik testler,fetal MRG ve multidisipliner perinatoloji konseyleri tanı sürecinde belirleyici rol oynar.
35 yaşında IVF gebelik,G1P0 hasta 21+1 haftada refere edildi.Ultrasonografide böbreklerin pelviste füzyone olduğu,parankimlerin hiperekojen ve renal pelvislerin belirgin olduğu izlendi(Figür1).Ayrıca tek umblikal arter ve hiperekojen barsak görüldü.Amniyosentez sonrası FISH, QF-PCR ve microarray normal sonuçlandı, fetal cinsiyet kromozomu XX olarak geldi.WES bakılmasını aile istemedi.
24+6 haftada sol renal pelvis AP çapı 7,1 mm ölçüldü.Fetal kalpte sağ ventrikül sola göre küçük, duvarlarda ekojenite artışı ve triküspit regürjitasyon izlendi; pulmoner stenoz açısından yakın takip önerildi.

29.haftada sol böbrek superiorunda 24x20 mm düzgün sınırlı kitle(Figür2),dilate rektum(Figür3),kliteromegali ve labial hipertrofi dikkati çekti.Ayrıcı tanıda adrenal kaynaklı androjen salgılayan kitle, konjenital adrenal hiperplazi, nöroblastom, virilizan tümörler ve renal kitle/kanama düşünüldü.Fetal MRG'de at nalı böbrek, sol megaüreter ve batın orta hatta kistik nodüler lezyon görüldü. Terminasyon istemeyen gebenin takipleri yapılarak 38+0 haftada malprezentasyon ve azalmış amniyon sıvı volümü nedeniyle sezaryen ile doğumu gerçekleştirildi. 3080 gr/ 48 cm ölçülerinde 1. ve 5. dakika APGAR skoru 8 ve 9 olan kız bebek doğurtuldu.İnspeksiyonda kliteromegali,labial hipertrofi izlendi(Figür4).Postnatal 2. günde anal atrezi nedeniyle kolostomi yapıldı. Yüksek üre, kreatinin ve potasyum değerleri saptanan yenidoğana böbrek yetmezliği nedeniyle periton diyalizi başlandı. At nalı böbrek ve renal displaziye eşlik eden kardiyak defekt, anal atrezi ve ambigius genitalya, kompleks anomalilerde multidisipliner yaklaşım gerekliliğini göstermektedir.Olgu,prenatal tanının ve fetal MRG'nin doğum sonrası yönetim için yol gösterici rolünü vurgulamaktadır.

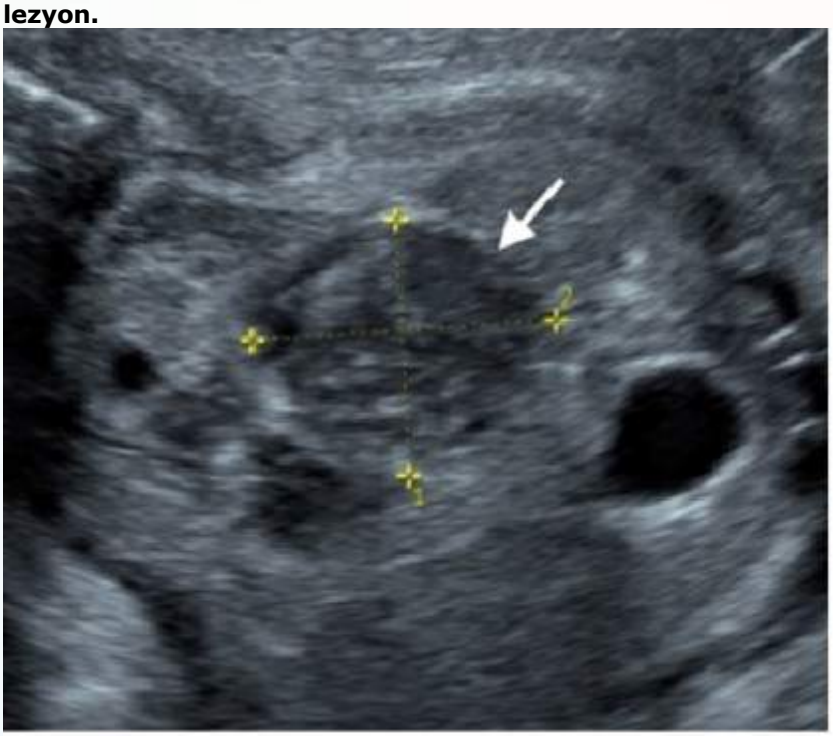
Anahtar Kelimeler: Anogenital malformasyon, Ektopik displastik böbrekler, Kliteromegali

Figür 1: 21+1 gebelik haftasında fetal böbreklerin pelviste füzyone olduğu, parankimlerinin displastik, hiperekojen ve renal pelvislerin belirgin olduğu izlendi.



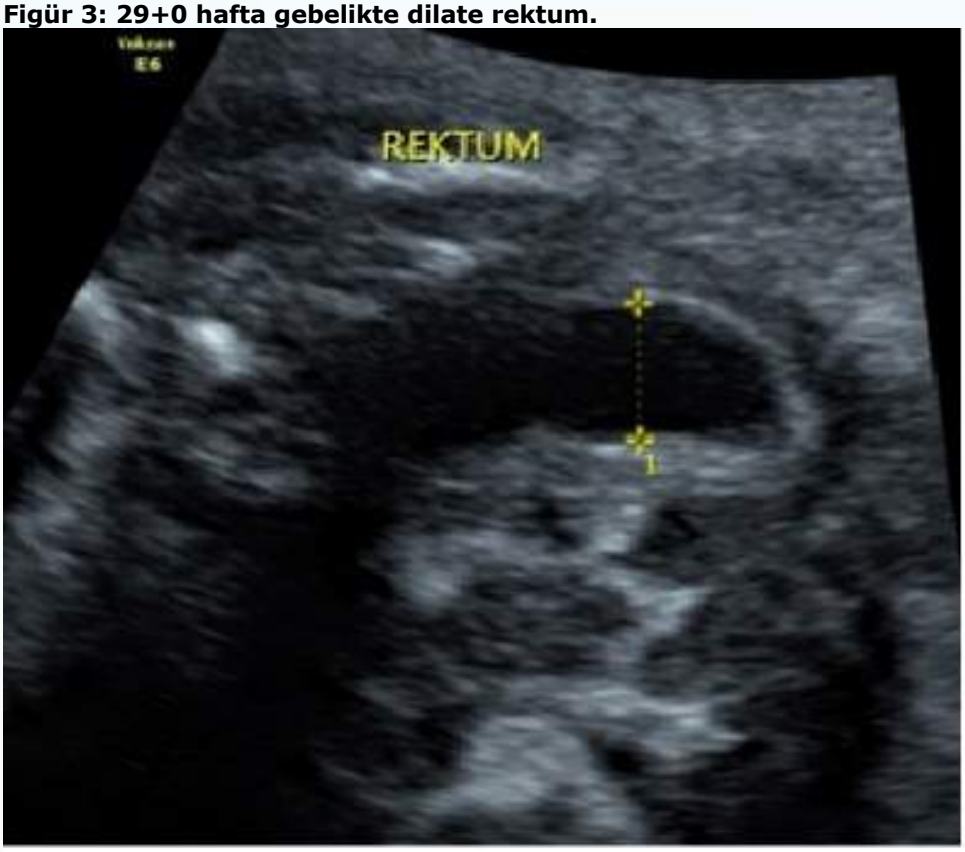
21+1 gebelik haftasında fetal böbreklerin pelviste füzyone olduğu, parankimlerinin displastik,hiperekojen ve renal pelvislerin belirgin olduğu izlendi. G: fetal mide, RK: sağ böbrek, LK: sol böbrek, B:mesane

Figür 2: 29+0 hafta gebelikte sol böbrek superiorunda 24x20mm düzgün sınırlı kitlesel lezyon.



29+0 hafta gebelikte sol böbrek superiorunda 24x20mm düzgün sınırlı kitlesel lezyon (ok işareti: kitlesel lezyon).

Figür 3: 29+0 hafta gebelikte dilate rektum.



29+0 hafta gebelikte dilate rektum.

Figür 4: Doğum sonrası kliteromegali ve labial hipertrofi görüntüsü.

Doğum sonrası kliteromegali ve labial hipertrofi görüntüsü.

PS-027

Comparison of the effects of Thymoquinone and Insulin in experimental gestational diabetic rats

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OBJECTIVE: This study aimed to compare the effects of thymoquinone and insulin on leptin, vascular endothelial growth factor (VEGF), blood glucose, and histopathological changes in the uterus and pancreas of experimental gestational diabetic rats.

MATERIALS-METHODS: Forty female rats were included in this experimental study. Group K1 (n=8) consisted of non-pregnant rats, while Group K2 (n=8) included healthy pregnant controls. Gestational diabetes mellitus (GDM) was induced in 24 pregnant rats via streptozotocin injection. These were further divided into three groups: GDM (n=8), GDM+TQ (n=8; treated with thymoquinone), and GDM+Ins (n=8; treated with insulin). Body weight and fasting blood glucose were measured at confirmation of pregnancy, day 6, and day 21. Uterine and pancreatic tissues were collected on day 21. Statistical significance was set at $p<0.05$.

RESULTS: In GDM rats, thymoquinone demonstrated antihyperglycemic effects comparable to insulin, with statistically significant reductions in fasting glucose. VEGF levels were decreased and leptin levels elevated in GDM rats compared with healthy pregnant controls. Both thymoquinone and insulin showed similar trends of increasing VEGF and decreasing leptin in GDM rats, though without statistical significance.

CONCLUSION: Thymoquinone exhibited an antihyperglycemic effect comparable to insulin, but no significant impact on leptin or VEGF levels was observed. While these findings suggest potential therapeutic properties, further studies are required to clarify its role as a candidate treatment for gestational diabetes mellitus

Keywords: gestational diabetic rat, streptozotocin, thymoquinone

PS-028

Fetal Adrenal Mass

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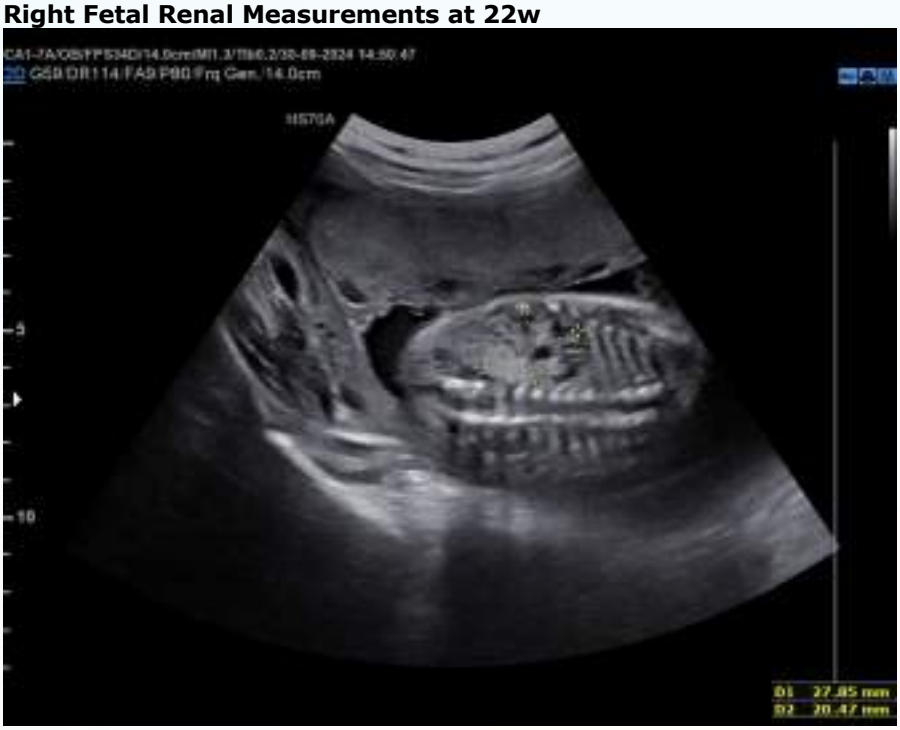
Introduction and OBJECTIVE: This case report presents the prenatal diagnosis and postnatal management of a solid-cystic mass located in the right adrenal region, ultimately diagnosed as fetal neuroblastoma.

Case Presentation: A 24-year-old primigravida woman with no known systemic disease was referred to our center at 22 weeks and 2 days of gestation due to a suspicious cystic lesion in the fetal liver/right adrenal region detected by ultrasound. Serial ultrasonographic follow-up revealed a mildly progressive solid-cystic mass. Fetal MRI confirmed the diagnosis in favor of neuroblastoma. Apart from polyhydramnios, no additional fetal anomaly was identified. At 39 weeks and 6 days, a 3080 g female infant was delivered via spontaneous vaginal birth. Postnatal imaging showed a heterogeneous, contrast-enhancing solid-cystic lesion in the right adrenal gland. Neuron-specific enolase (NSE) was elevated at 77.1 ng/mL. Due to the potential for spontaneous regression, the patient was managed conservatively by pediatric hematology-oncology. On follow-up, NSE level regressed to 26 ng/mL as expected.

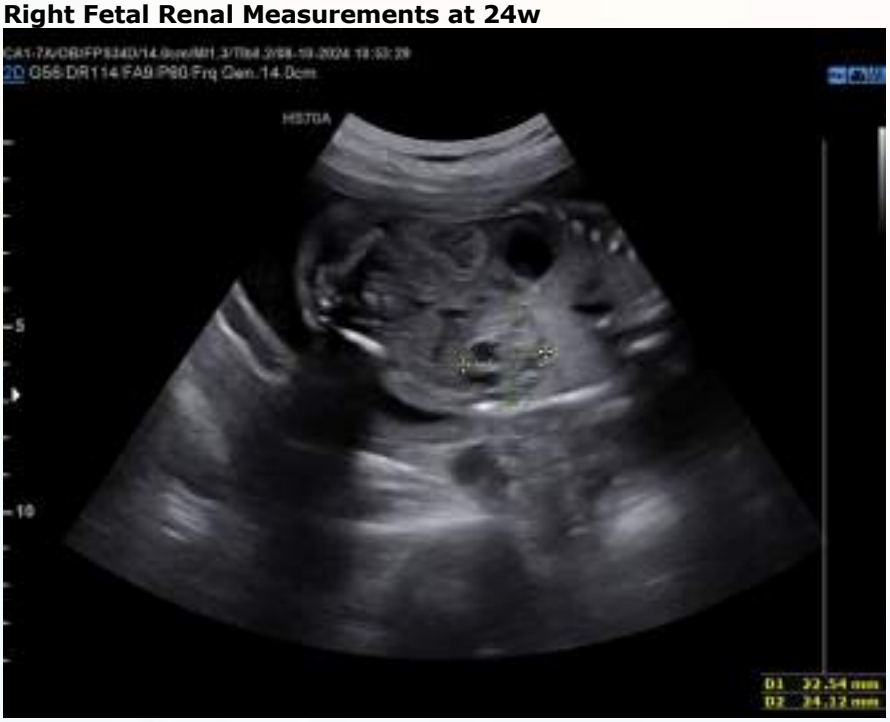
CONCLUSION: Fetal neuroblastoma is the most common adrenal neoplasm diagnosed prenatally, with ultrasonography being the primary diagnostic tool. As in this case, MRI significantly contributes to differential diagnosis and assessment of tumor characteristics. Spontaneous regression is a notable feature of neuroblastoma, especially in cases diagnosed during the prenatal or early infantile period. Therefore, conservative management can be a viable option in select cases, avoiding unnecessary invasive procedures. Fetal neuroblastoma is a condition that requires a multidisciplinary approach, with diagnosis facilitated by advanced prenatal imaging and follow-up.

Keywords: Fetal MRI, Neuroblastoma, Prenatal Diagnosis, Screening Ultrasonography.

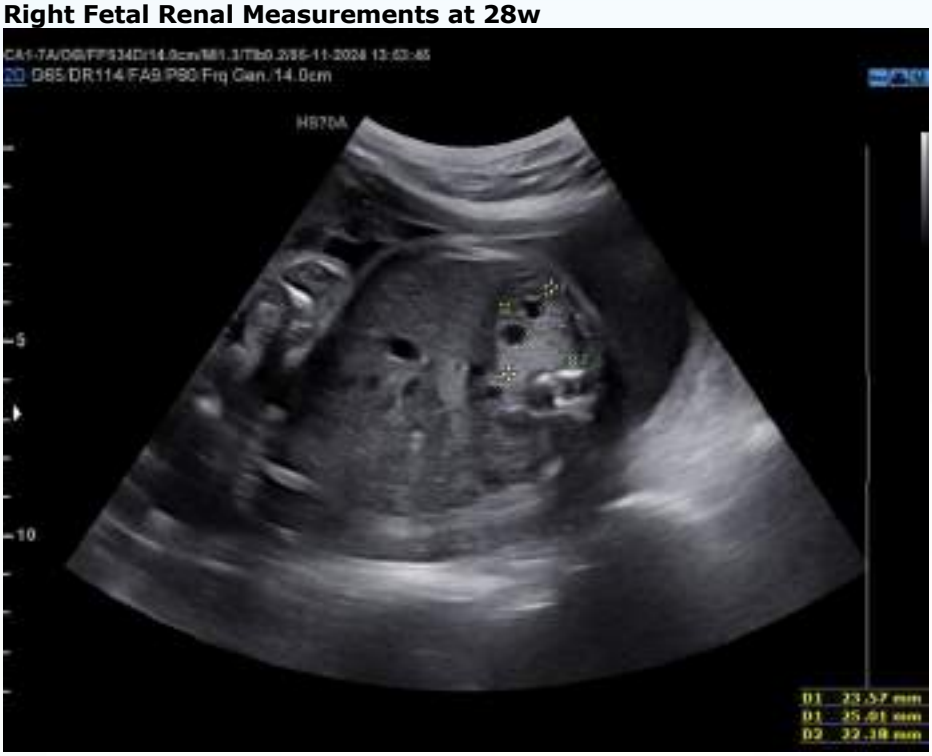
Right Fetal Renal Measurements at 22w



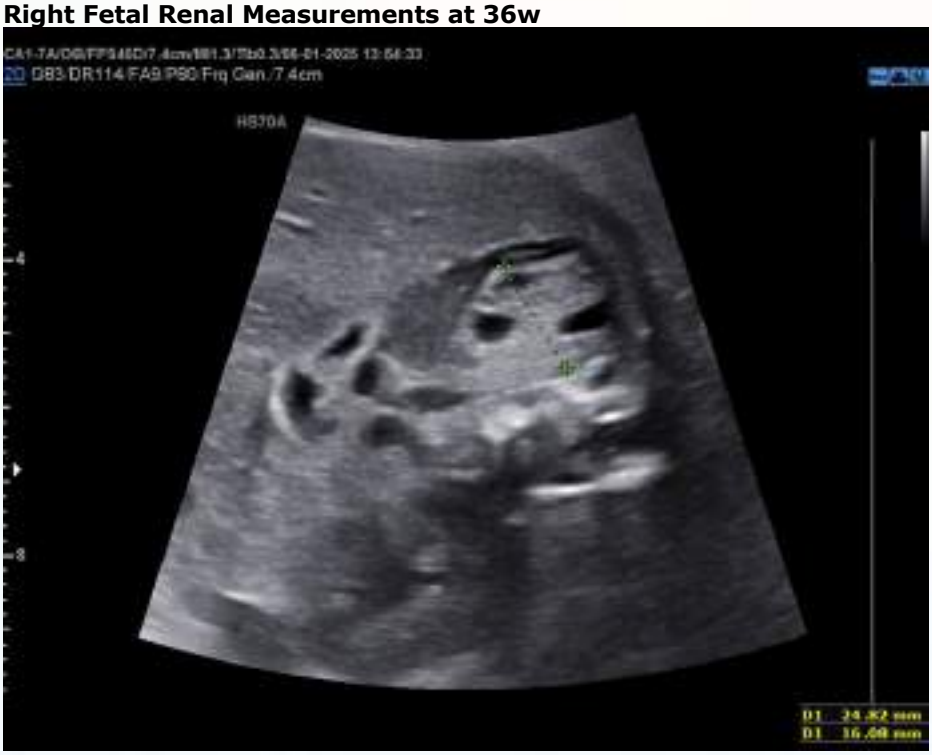
Right Fetal Renal Measurements at 24w



Right Fetal Renal Measurements at 28w



Right Fetal Renal Measurements at 36w



PS-029

Retinoic Acid Embryopathy: A Case Report

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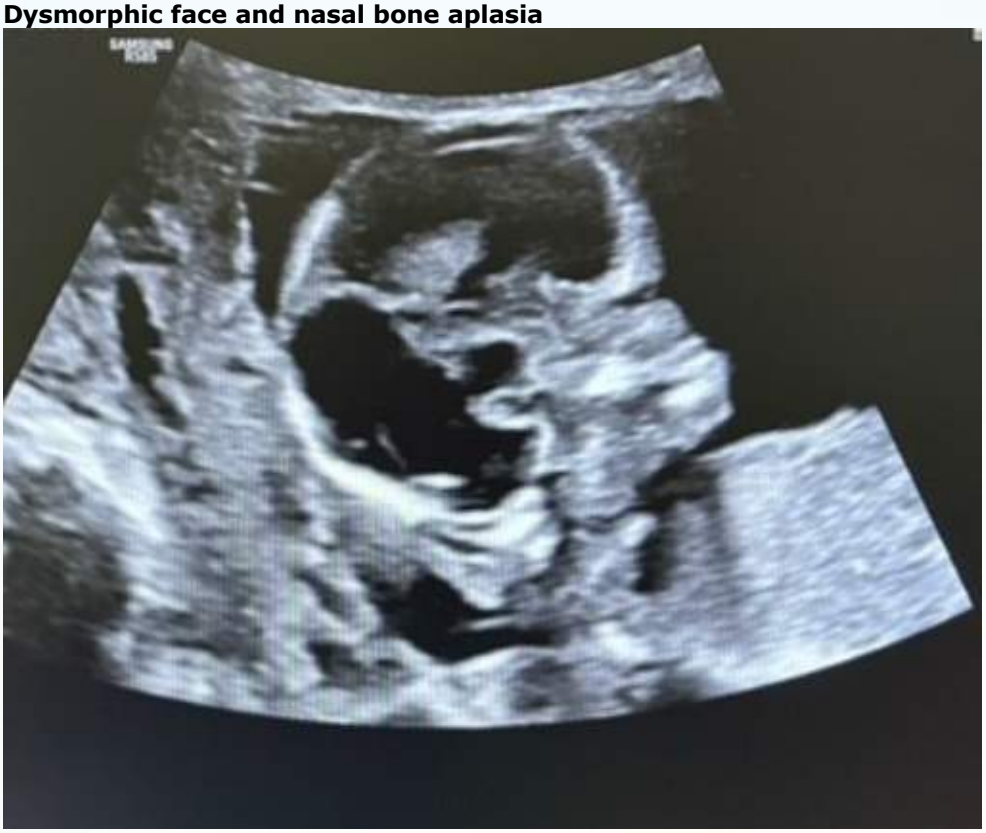
OBJECTIVE: This case report aims to present the characteristic teratogenic effects and clinical management of a pregnancy inadvertently exposed to isotretinoin, highlighting the critical importance of strict pregnancy prevention measures.

Case A 21-year-old primigravida was referred at 16 weeks of gestation following abnormal antenatal screening. She had inadvertently continued oral isotretinoin for severe acne throughout early pregnancy. A detailed ultrasound revealed a classic pattern of malformations consistent with retinoic acid embryopathy, including skull deformities, facial abnormalities, hydrocephalus, and a complex cardiac defect (atrioventricular septal defect). After extensive multidisciplinary counselling regarding the severe prognosis and significant developmental disabilities associated with these findings, the patient opted for pregnancy termination. Postmortem examination confirmed the extensive craniofacial dysmorphism and complex cardiac pathology, providing definitive confirmation of the diagnosis.

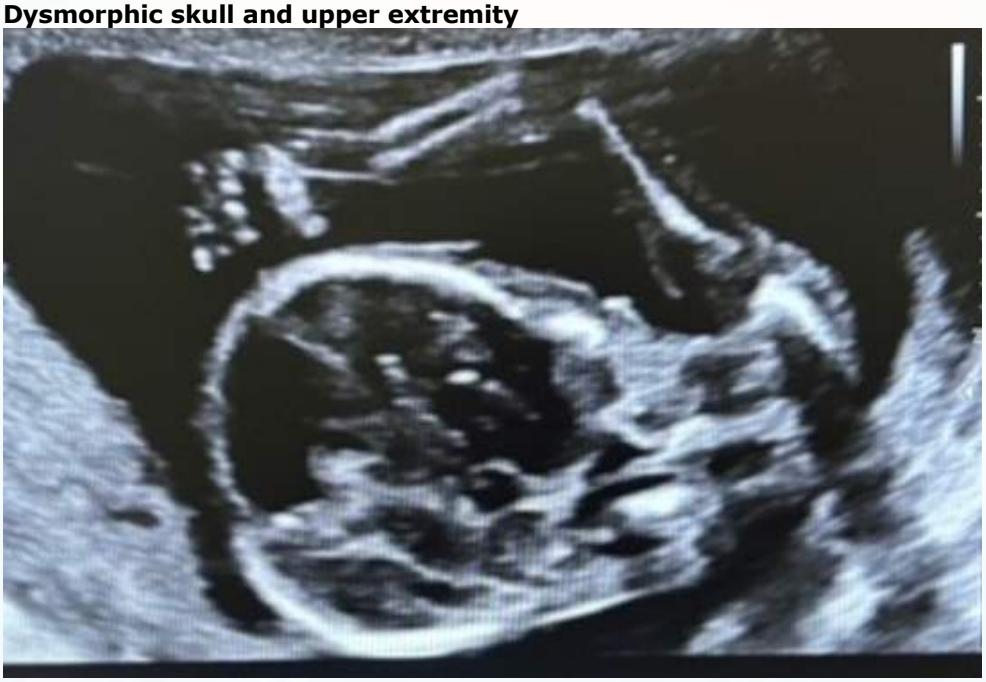
CONCLUSION: This case underscores the high teratogenic risk of isotretinoin and the devastating, multi-system fetal anomalies it can cause. It reinforces the absolute necessity for robust pregnancy prevention programmes, including mandatory pre-treatment counselling, reliable contraception, and monthly pregnancy testing for all women of reproductive age prescribed this medication. Strict adherence to these protocols is essential to prevent such outcomes.

Keywords: Embryopathy, Pregnancy, Retinoic Acid

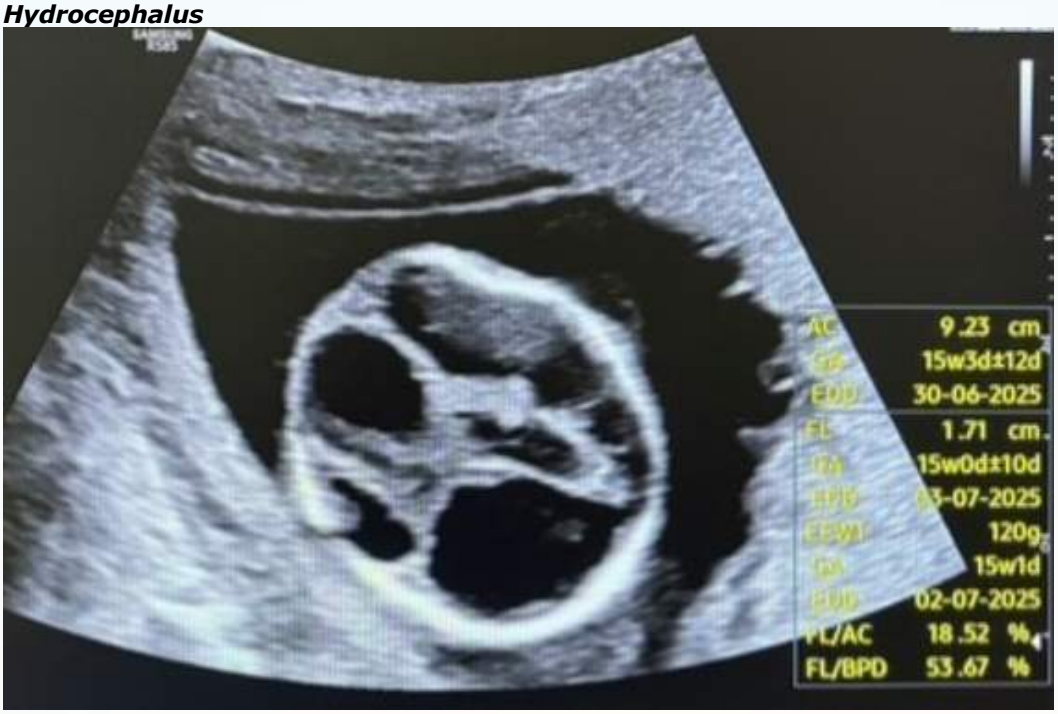
Dysmorphic face and nasal bone aplasia



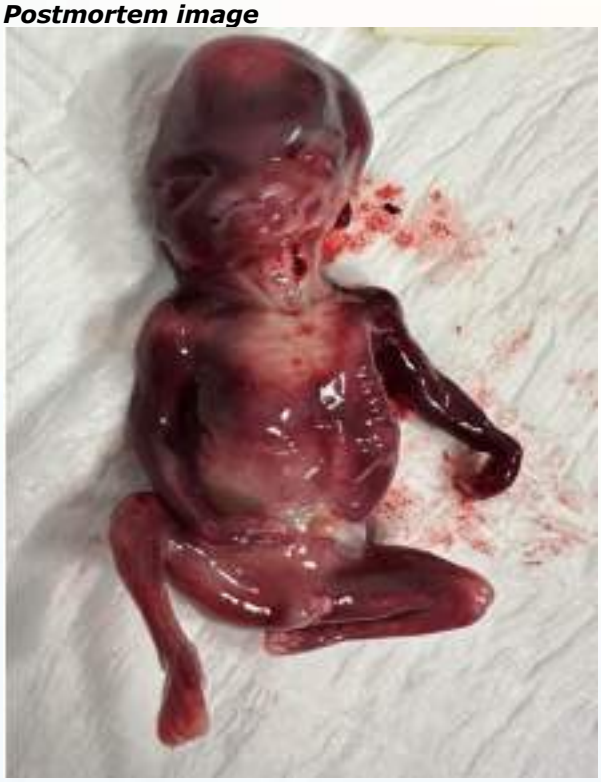
Dysmorphic skull and upper extremity



Hydrocephalus



Postmortem image



Dysmorphic face and skull, microphthalmia, dysmorphic upper extremities

Türkiye Maternal Fetal Tıp ve Perinatoloji Derneği
Ultrasonografi Kongresi
29 Ekim - 1 Kasım 2025
Renaissance Polat İstanbul Hotel - Yeşilköy

PS-030

İkinci Trimester Ultrasonografisinde Pulmoner Stenoz, İleri Derece Triküspit Regürjitasyonu ve Endokardial Fibroelastozis ile Seyreden Fetal Olgu

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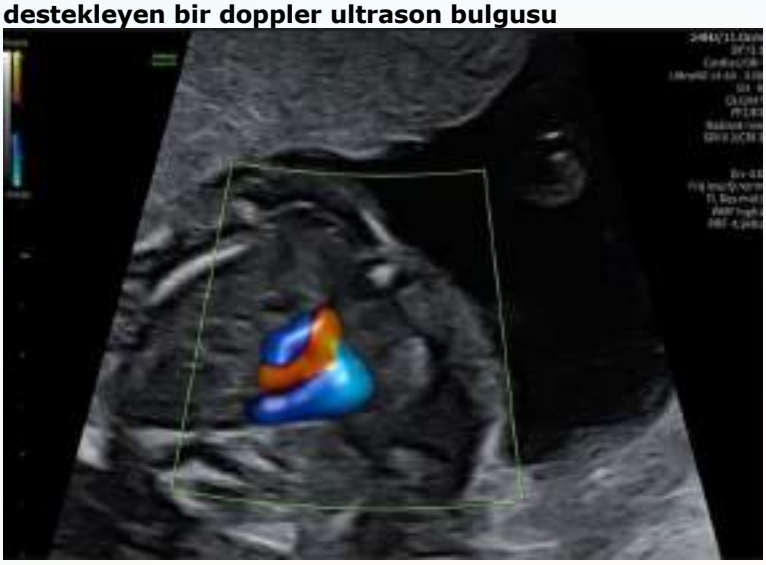
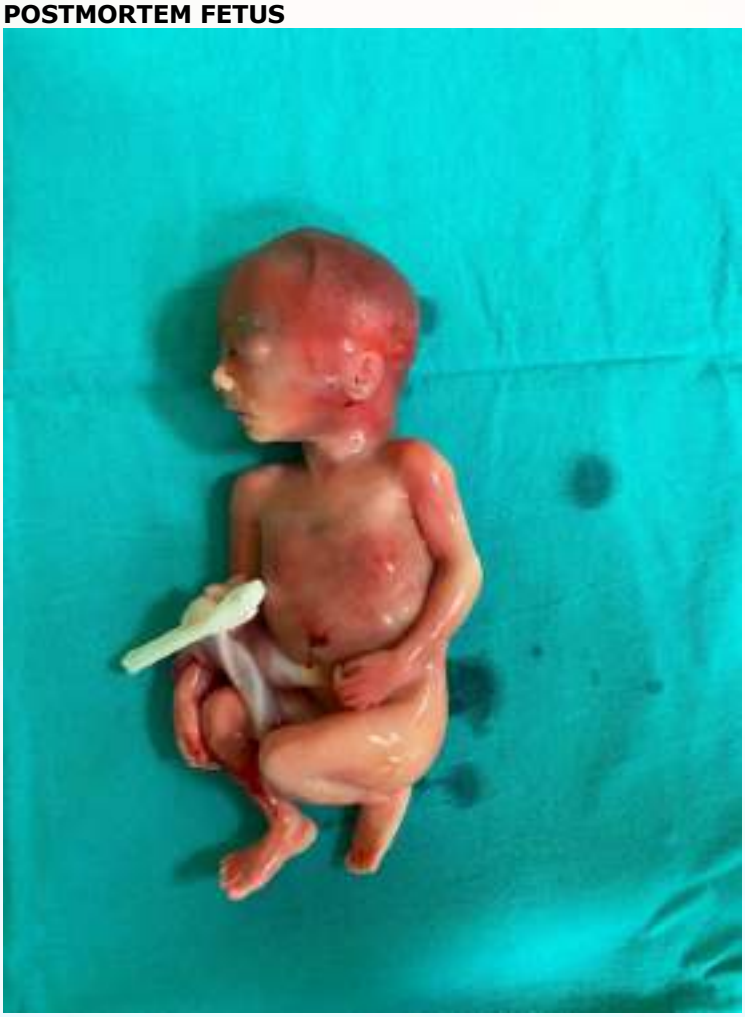
Otuz bir yaşında, gravida 1 parite 0 olan hasta son adet tarihine göre 20 hafta 4 günlük gebeliğiyle kliniğimize başvurdu. Maternal hematolojik ve biyokimyasal değerlerinde ve fizik muayenesinde herhangi bir anormallik saptanmadı. Maternal kanda yapılan fetal DNA aracılığı ile Down Sendromu, Trizomi 13, Trizomi 18 taraması negatif (Low risk, fetal fraksiyon % 5) bulunmuştur. Ultrasonografide intrauterin fetal kalp atımı görülen baş prezentasyonda gebelik izlendi. Yapılan fetal ekokardiyografide, Ventriküler Septal Defektin (VSD) eşlik etmediği ciddi pulmoner stenoz saptandı. Pulmoner kapak hizasında belirgin darlık ve ileri-geri akım izlendi. Sağ ventrikülün sol ventriküle göre küçük olduğu, sağ ventrikül duvarında ekojenite artışı (endokardial fibroelastozis ile uyumlu) gözlendi. Ayrıca triküspit kapakta atriyum duvarına kadar ulaşan şiddetli triküspit regürjitasyonu tespit edildi. Hasta son adet tarihine göre 20 hafta 5 günlük gebe iken İstanbul Tıp Fakültesi Perinatoloji-Kardiyoloji konseyinde değerlendirilerek amniyosentez işlemi gerçekleştirildi. Amniyosentez sonucunda uzun süreli hücre kültürü ve mikroarray sonucu normal saptandı. İleri tarih için tüm ekzom analizi(WES) planlandı. Gebeliğinin 21. haftasında aileye prognoz hakkında danışmanlık verildi ve tıbbi terminasyon kararı alındı. Fetusa fetosit amacıyla KCl intrakardiyak uygulandı ve ardından misoprostol protokolü ile gebelik sonlandırıldı. Pulmoner stenoz ve ileri triküspit regürjitasyonu ile birlikte endokardial fibroelastozis görülmesi, kötü fetal prognoz ile ilişkili ciddi bir kardiyak anomalidir. Prenatal dönemde yapılan detaylı fetal ekokardiyografi ile erken tanı konulması, genetik analizlerin planlanması ve aileye prognoz hakkında danışmanlık verilmesi açısından kritik önem taşır. Bu olgu, fetal kardiyak anomalilerin multidisipliner değerlendirme ve bireyselleştirilmiş yönetim geolgu, fetal kardiyak anomalilerin multidisipliner değerlendirme ve bireyselleştirilmiş yönetim gerektirdiğini vurgulamaktadır.rektirdiğini vurgulamaktadır.

Anahtar Kelimeler: Pulmoner stenoz, triküspit regürjitasyonu, endokardial fibroelastozis, fetal ekokardiyografi

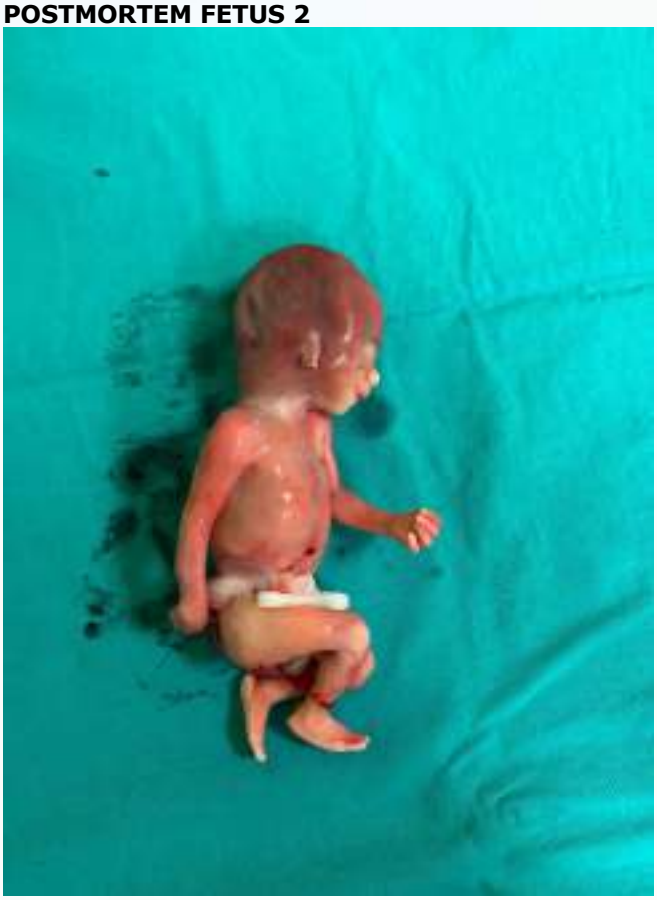
Ciddi triküspit yetersizliği veya pulmoner stenoza bağlı ters akım (regürjitasyon) varlığını destekleyen bir doppler ultrason bulgusu



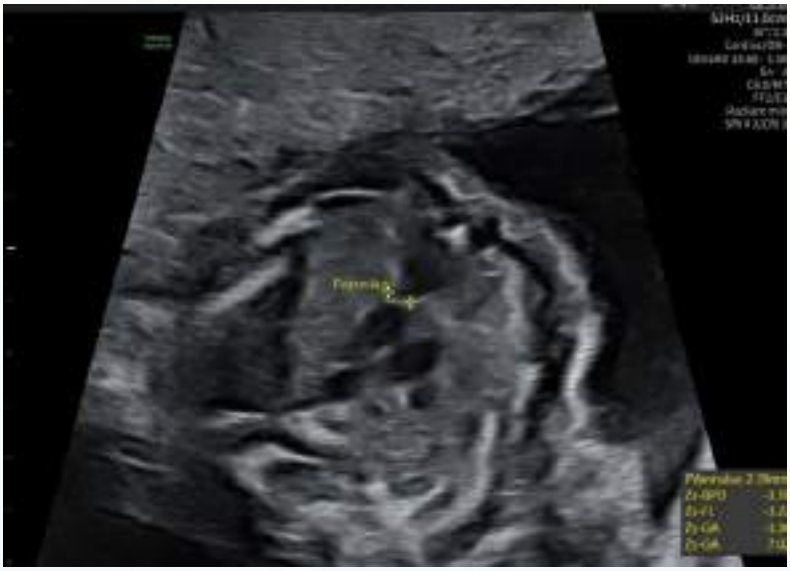
POSTMORTEM FETUS



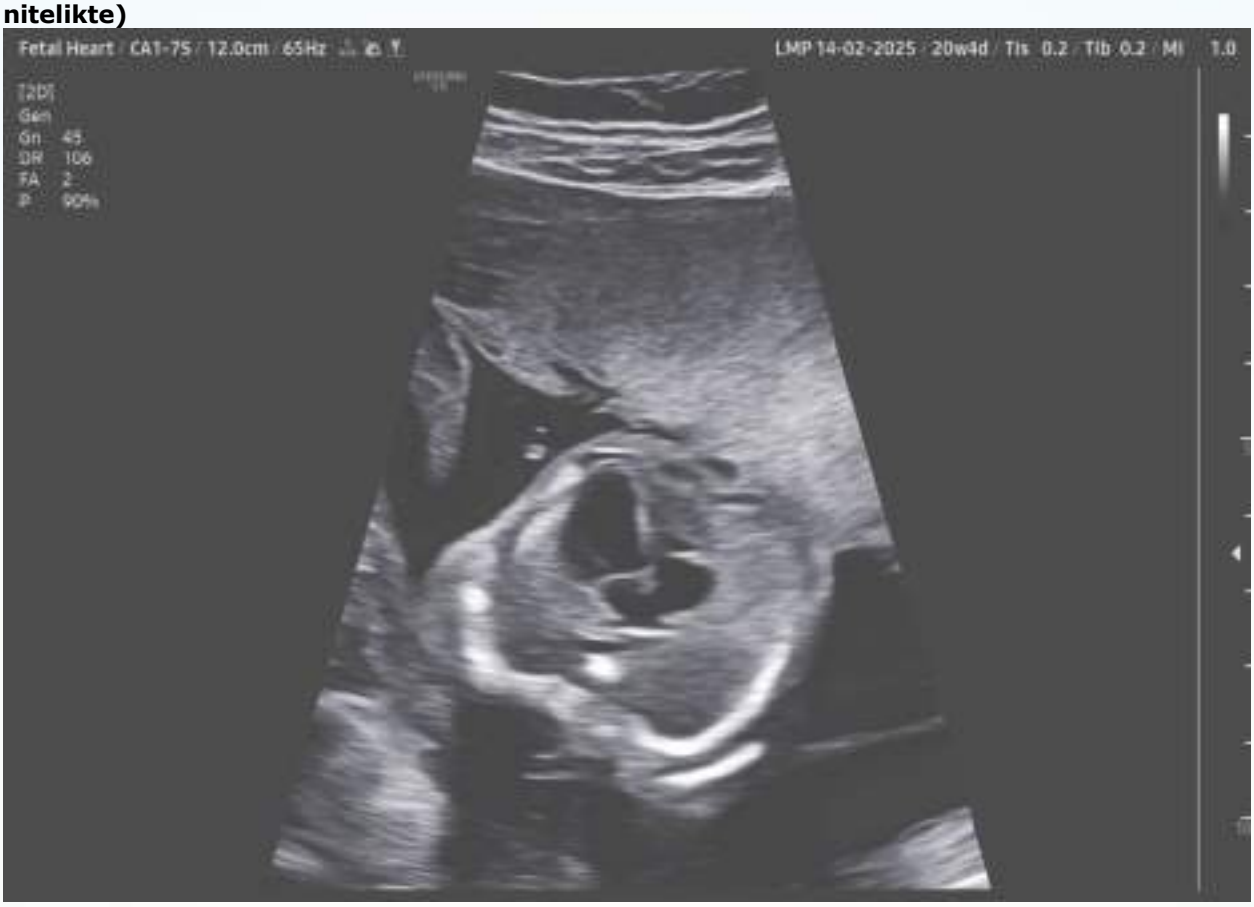
POSTMORTEM FETUS 2



Pulmoner Stenoz



Sağ ventrikül duvarında artmış ekojenite (endokardial fibroelastozis bulgusunu destekler nitelikte)



Türkiye Maternal Fetal Tıp ve Perinatoloji Derneği
Ultrasonografi Kongresi
29 Ekim - 1 Kasım 2025
Renaissance Polat İstanbul Hotel - Yeşilköy

PS-031

A Rare Case: Maternal Pelvic Spleen

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Introduction and OBJECTIVE: Ectopic spleen is a rare clinical malformation resulting from the absence or laxity of gastrosplenic and splenorenal ligaments, causing the spleen to be located outside its normal anatomical position in the left upper quadrant. This condition is 13 times more common in women and can lead to pregnancy and delivery complications. This case highlights the importance of recognizing ectopic spleen as a potential source of obstetric complications in women of reproductive age.

Case Presentation: We report the case of a 32-year-old woman, gravida 4 parity 3, at 34 weeks and 1 day of gestation, who came to our emergency department with pain. The patient had a previous history of known pelvic spleen. Prior abdominal computed tomography revealed a 164×114×104 mm wandering spleen extending from the umbilical level inferiorly to the posterosuperior aspect of the uterus, with a characteristic whirlpool pattern of splenic vessels. The patient underwent cesarean section under the indication of painful previous cesarean section. During surgery, an ectopic spleen was identified in the Douglas pouch, adherent to the posterosuperior aspect of the uterus. After the suturizing the uterus, active bleeding was observed from the ectopic spleen bed. General surgery was consulted intraoperatively. The adhesions between the uterus and ectopic spleen were carefully ligated. Hemostasis was achieved through cauterization of the bleeding areas on the splenic surface.

CONCLUSION: Ectopic spleen can present significant challenges during cesarean delivery. Preoperative awareness of this condition and multidisciplinary intraoperative management are essential for successful outcomes.

Keywords: cesarean section, maternal pelvic spleen, wandering spleen.

Douglas Pouch, After Ligating the Invasive Vessels

Douglas Pouch, Spleen Invasion Area to Posterior of the Uterus

PS-032

Heterotopik sezaryen skar gebeliği: Olgu sunumu

Fedi Ercan, Ebru Yaltı
Serbest Hekim, Aydın

AMAÇ: Heterotopik gebelik; eşzamanlı intrauterin gebelik ile ekstrauterin yerleşimli ikinci bir gebeliğin birlikteliğidir. Sezaryen skar gebeliği (SSG) heterotopik olguların nadir bir alt tipidir. Bu olgu, iki kez sezaryen öyküsü olan, son adet tarihine göre 5 haftalık, 34 yaşındaki hastamızda saptanan intrauterin + skar yerleşimli heterotopik gebeliğin tanı ve yönetimini sunmaktadır.

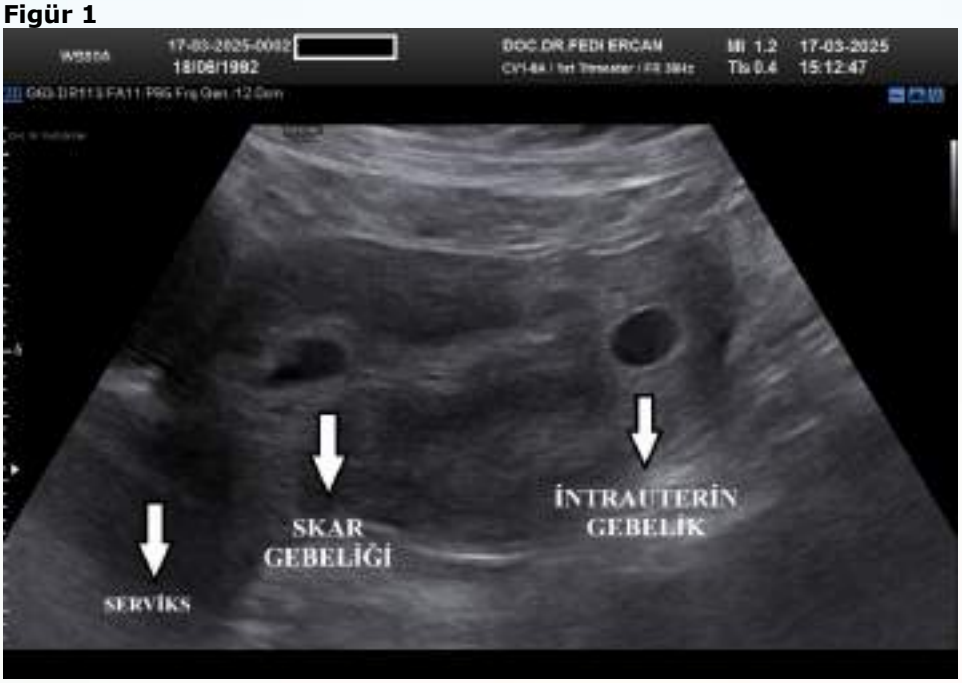
OLGU: Adet rötarı ile başvuran 34 yaşında daha önce 2 alt segment transvers kesi ile sezaryen öyküsü olan hastanın transabdominal ve transvajinal ultrasonunda uterin kavitede gestasyonel kese ile alt uterin segmente, önceki histerotomi hattına komşu ikinci bir kese izlendi (Şekil 1 ve 2), her iki görüntüde intrauterin ve skar gebelikleri oklarla işaretlendi. Heterotopik SSG düşünüldü. Hastaya olası komplikasyonlar (kanama, uterin rüptür, gelecekteki fertiliteye etkileri) anlatılarak tedavi yaklaşımları tartışıldı.

Yönetim: Hastamız gebeliğini sonlandırma isteğini bildirdi. Hazırlık sonrası, ultrason eşliğinde, hemostatik önlemlerle, hem intrauterin hem de skar yerleşimli gebelik aynı seansta vakum aspirasyon (küretaj) ile sonlandırıldı. İşlem komplikasyonsuz seyretti; postoperatif izlemde hemoglobin stabil, kanama minimaldi.

SONUÇ: Sezaryen skar alanında implantasyonun eşlik ettiği çok erken heterotopik gebelik, dikkatli bir ultrason muayenesi ile saptanabilir. Yönetim, hastanın çocuk istemi ve olası riskler tartışılarak bireyselleştirilmelidir. Hastamızda, hastanın tercihi doğrultusunda eş zamanlı küretaj güvenli biçimde uygulanmış, majör komplikasyon gelişmemiştir. Bu olgu, erken tanının danışmanlık kalitesini ve komplikasyonların önlenmesini kolaylaştırdığını; ayrıca SSG'de karar verirken görüntülemenin yönlendirici rolünü vurgular. Literatürde heterotopik gebeliğin spontan konsepsiyonlarda çok düşük sıklıkta bildirildiği ve ünite bazında standart yönetim kılavuzlarının sınırlı olduğu dikkate alındığında, olgumuzun erken dönemde tanınması ve güvenli sonlandırılması önemlidir.

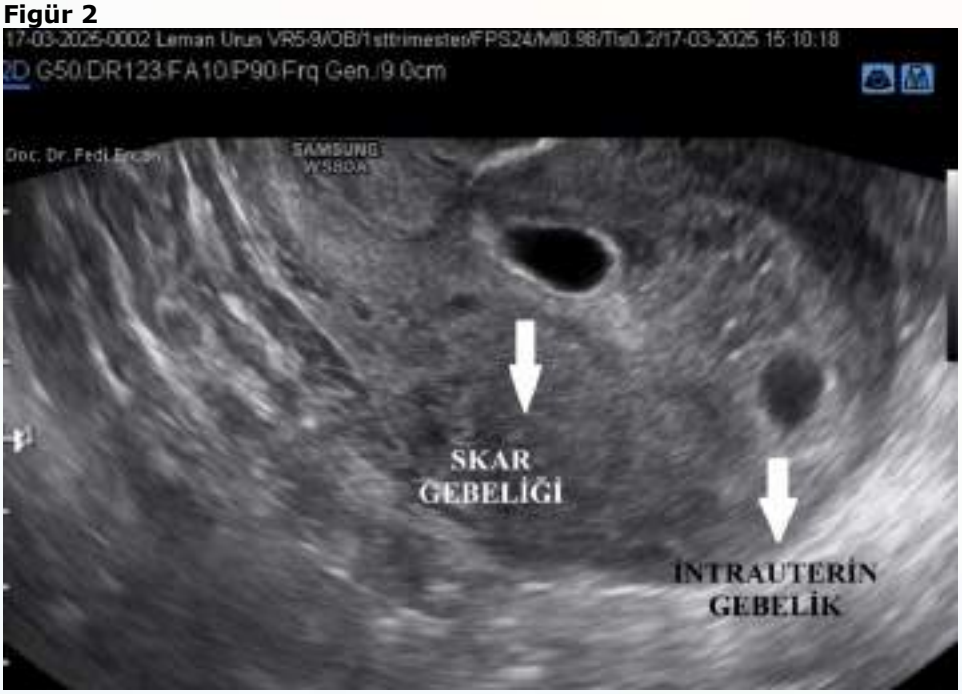
Anahtar Kelimeler: Ektopik gebelik, heterotopik gebelik, sezaryen skar gebeliği

Figür 1



Transabdominal ultrason incelemesinde servikse ve skar ile intrauterin gebelik keselerine ait görüntüler oklarla işaretlenerek gösterilmiştir.

Figür 2



Transvajinal ultrason incelemesinde sezaryen skar gebeliği ile intrauterin gebelik keselerine ait görüntüler oklarla işaretlenerek gösterilmiştir.

PS-033

A Case of Harlequin Ichthyosis on Prenatal Ultrasonography

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INTRODUCTION: Disorders of keratinization (ichthyoses) are a group of inherited diseases characterized clinically by dry, thick, and scaly skin, and histopathologically by hyperkeratosis. Harlequin ichthyosis is one of the most severe forms of congenital ichthyosis. It is inherited in an autosomal recessive manner and is extremely rare.

Case Presentation A 29-year-old woman, gravida 6, parity 4, abortus 1, was referred to our perinatology clinic at 30 weeks of gestation. Ultrasonographic examination revealed intrauterine growth restriction, reduced amniotic fluid and absent end-diastolic flow in the umbilical artery. The amniotic fluid appeared echogenic. Fetal biometry was consistent with gestational age. Based on these findings, the case was closely monitored. Delivery occurred at 34 weeks of gestation. Postnatal examination of the newborn revealed diffuse skin thickening, hyperkeratotic plaques, and a typical appearance of ichthyosis.

Discussion: Prenatal diagnosis of ichthyosis is extremely challenging. According to the literature, increased echogenicity of the amniotic fluid and specific fetal skin and facial findings may aid in diagnosis. In our case, reduced and echogenic amniotic fluid raised prenatal suspicion, which was later confirmed postnatally.

CONCLUSION: Prenatal ultrasonography can provide valuable clues for the early detection of rare genetic skin disorders such as ichthyosis. Our case highlights the importance of careful ultrasonographic evaluation in such conditions.

References

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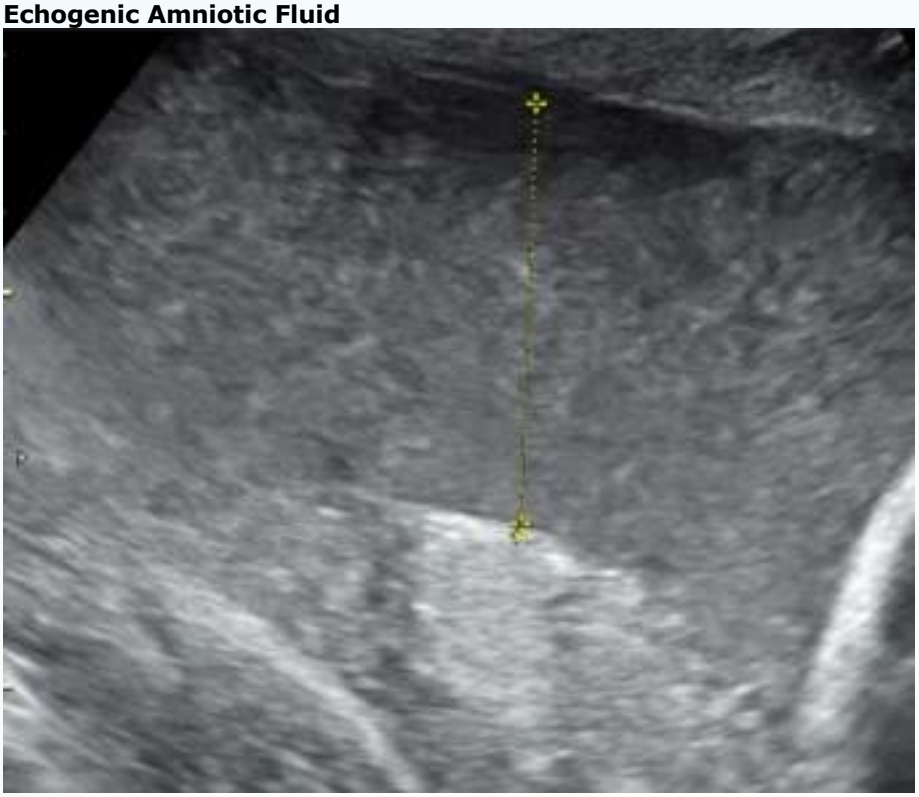
2. Briceag I, et al. Prenatal diagnosis of Harlequin ichthyosis by ultrasound and genetic testing. BMC Med Imaging.2021;21:117. doi:10.1186/s12880-021-00586-4

Keywords: Keywords Prenatal ultrasonography, ichthyosis, Harlequin ichthyosis

Echogenic Amniotic Fluid



Harlequin Ichthyosis-1

Harlequin Ichthyosis-2

Harlequin Ichthyosis-3

PS-034

Beyond The Neural Tube: Craniorachischisis With Omphalocele And Complex Cardiac Anomalies

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Craniorachischisis is the most severe and rare form of neural tube defect, characterized by anencephaly with a contiguous open spinal lesion. This condition is uniformly lethal. While its occurrence is rare in itself, the combination with additional severe anomalies is extraordinary. We present a unique case of craniorachischisis coexisting with omphalocele and multiple complex cardiac malformations.

A 25-year-old woman, gravida 5, para 3, abortus 1, was referred at 18+2 weeks. Ultrasonography demonstrated craniorachischisis. Counseling regarding the uniformly poor prognosis was provided, and pregnancy termination was offered; but the couple initially opted to continue. At 21+4 weeks, further evaluation revealed an omphalocele and complex congenital heart disease characterized by pulmonary valve atresia with intact ventricular septum, right ventricular hypoplasia, severe tricuspid regurgitation, and abnormal sinusoidal flow patterns. After repeated counseling, termination was accepted at 23+5 weeks Molecular karyotyping revealed no chromosomal abnormalities.

Maternal history revealed no folic acid supplementation during pregnancy.. There was no consanguinity, diabetes, or smoking. Preconception labs demonstrated microcytic anemia (Hb 9.6 g/dL, MCV 63 fL) without megaloblastic features; B12 was normal and folate borderline low (5.09 ng/mL). Her three children were healthy.

This case highlights an extraordinary association of craniorachischisis, omphalocele, and complex cardiac malformations. Such combinations are exceptionally rare in the literature and suggest a profound disruption of early embryogenesis. Prenatal diagnosis, repeated multidisciplinary evaluation, and timely counseling are essential for informed parental decision-making and perinatal management in lethal and complex malformations.

Keywords: craniorachischisis, omphalocele, cardiac anomalies

Figure-1

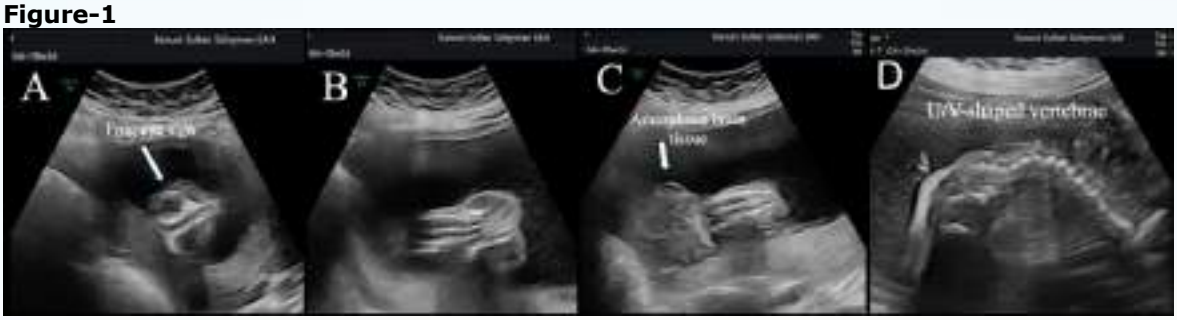

Figure-1 Second trimester ultrasound A; coronal section through fetal face showing frog-eye deformity and absence of calvaria B; Coronal view of the fetus showing total open spina bifida, from the cervical region to the sacral region of the spine. C; Sagittal view showing Residual disorganized brain tissue was observed floating in the amniotic fluid and connected to the abnormal spinal cord. D; Sagittal; view, the vertebral column appeared irregularly curved and discontinuous, consistent with severe open spinal dysraphism

Figure-2

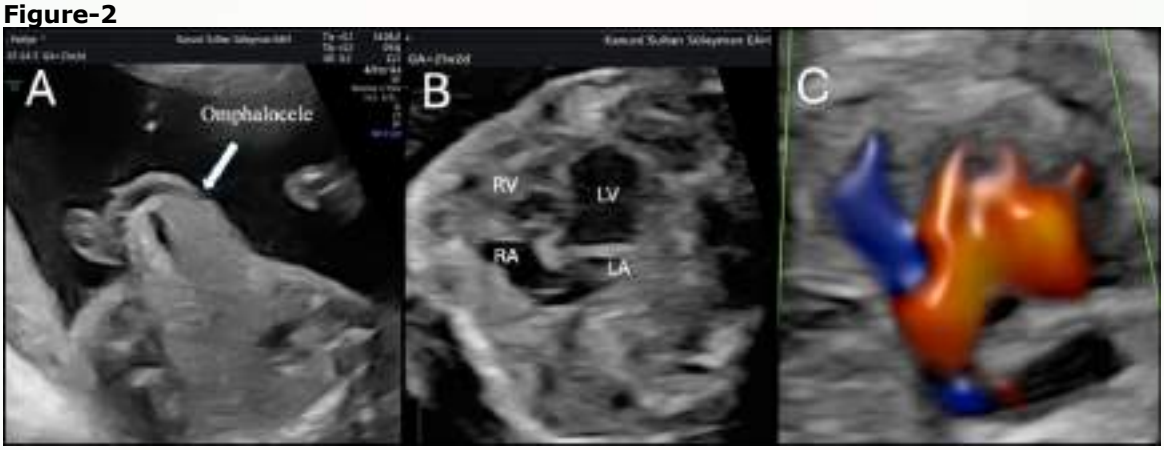

Figure-2 Second trimester ultrasound(21w+2) A; Axial ultrasound view showing a midline anterior abdominal wall defect containing liver and bowel loops within a membranous sac, consistent with omphalocele. B; Four-chamber ultrasound view demonstrating right ventricular hypoplasia and ventricular asymmetry C; Three-vessel Doppler view demonstrating reversed flow in the pulmonary artery, consistent with pulmonary valve atresia.

Figure-3

Figure 3 Macroscopic view of the fetus showing total absence of normal brain structures and a completely open spinal defect. Cranially, the fetus exhibits the characteristic frog-eye deformity, broad nasal bridge, low-set and abnormally shaped ears, prominent eyes, macroglossia, short neck, and skeletal malformations with vertebral defects.

PS-035

Erken Gebelik Haftasında Tanı Alan Sol Konjenital Diyafragma Hernisinde Prenatal Ultrasonografinin Klinik Yönetimdeki Rolü

Gözde Karataş[1], Şule Birol İnce[1], Recep Has[2]

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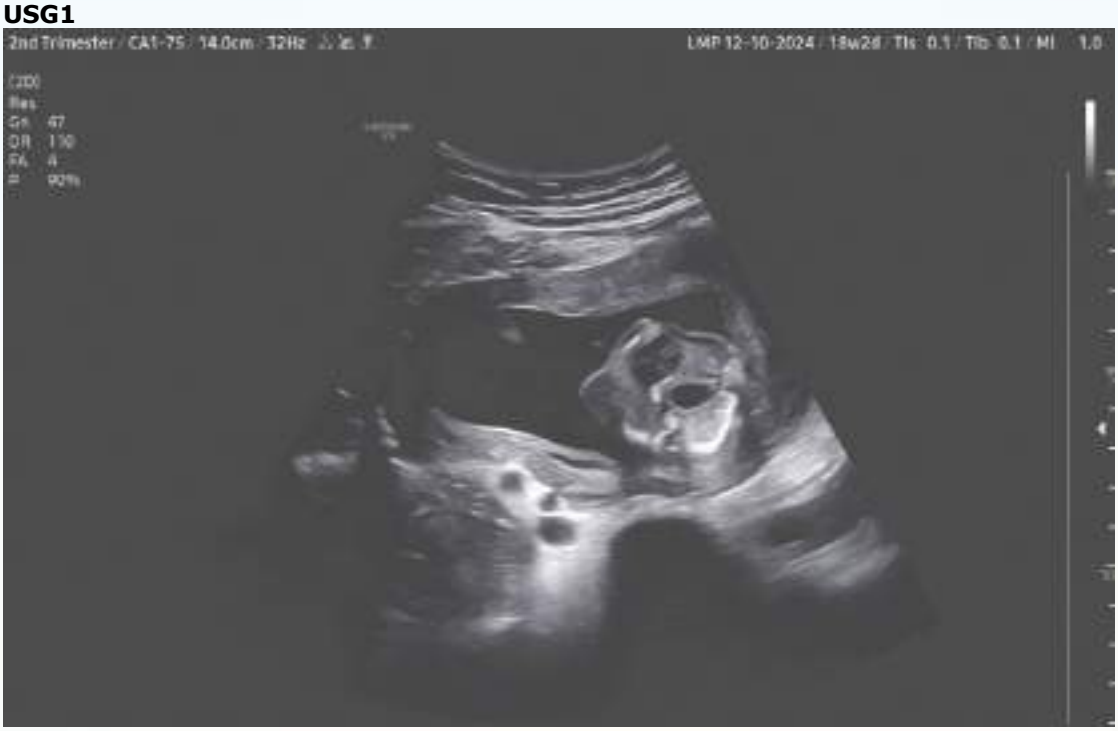
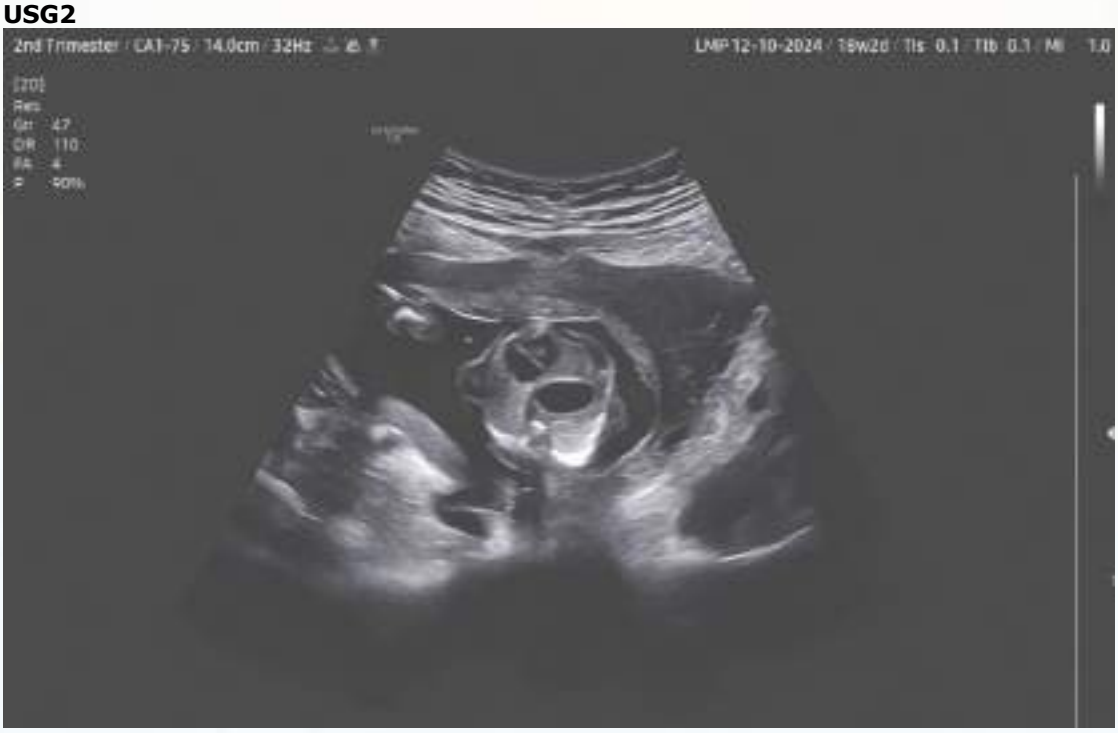
25 yaşında, ilk gebeliğinde erken dönem abortus, ikinci gebeliğinde şiddetli preeklampsi nedeniyle 33. haftada 1500 gr sezaryen doğum, neonatal exitus öyküsü bulunan, mevcut gebeliğinde coraspirin kullanan hasta; son adet tarihine(SAT) göre 16 hafta 5 günlük gebeliğiyle kliniğimize başvurdu. Hematolojik ve biyokimyasal değerlerinde ve fizik muayenesinde herhangi bir anormallik saptanmadı. Ultrasonografisinde intrauterin fetal kalp atışı olan tekil gebelik izlendi. Hastanın ilk trimester tarama testinde Trizomi 21 Riski: 1/35500, Yaş Riski: 1/1330, Trizomi 18 Riski: 1/1740 olarak görüldü. Hastanın SAT'a göre 16 hafta 5 günlük gebe iken yapılan ultrasonografide sol diyafragma hernisi görüldü, mide cebi toraks içerisinde izlendi, fetal kalbin sağa itildiği ve O/E LHR(Jani) %22 olarak hesaplandığı görüldü. Bu bulgular, prognozu kötü etkileyebilecek ciddi diyafragma hernisi lehine değerlendirildi. Hasta genetik danışmaya yönlendirildi. Hasta SAT'a göre 16 hafta 6 günlük gebe iken Perinatoloji konseyinde değerlendirilerek amniyosentez işlemi gerçekleştirildi. Amniyosentez sonucunda uzun süreli hücre kültürü ve mikroarray sonucunda uzun süreli hücre kültürü sonucu normal saptandı. İleri tarih için tüm ekzom analizi(WES) planlandı. Hastanın SAT'a göre 18 hafta 2 günlük gebe iken tekrarlanan değerlendirmesindeki bulgularla sol diyafragma hernisinin olumsuz prognozu hakkında aileye danışmanlık verildi ve tıbbi terminasyon kararı alındı. Misoprostol protokolüne göre indükte abort gerçekleşti. Postmortem incelemede sol diyafragma hernisi, fasiyal dismorfizm olarak değerlendirildi. Konjenital diyafragma hernisi, prenatal dönemde özellikle erken haftalarda yapılan detaylı ultrasonografik inceleme ile saptanabilir. Erken tanı, anomalinin ciddiyetini değerlendirme ve gebelik yönetimi hakkında bilinçli karar verme sürecini kolaylaştırır. Bu olguda olduğu gibi, kötü prognoz öngörülen durumlarda erken tanı, gebelik sonlandırılması da dahil olmak üzere uygun klinik yaklaşımın belirlenmesine katkı sağlamaktadır.

Anahtar Kelimeler: Konjenital diyafragma hernisi, prenatal tanı, ultrasonografi, fetal prognoz

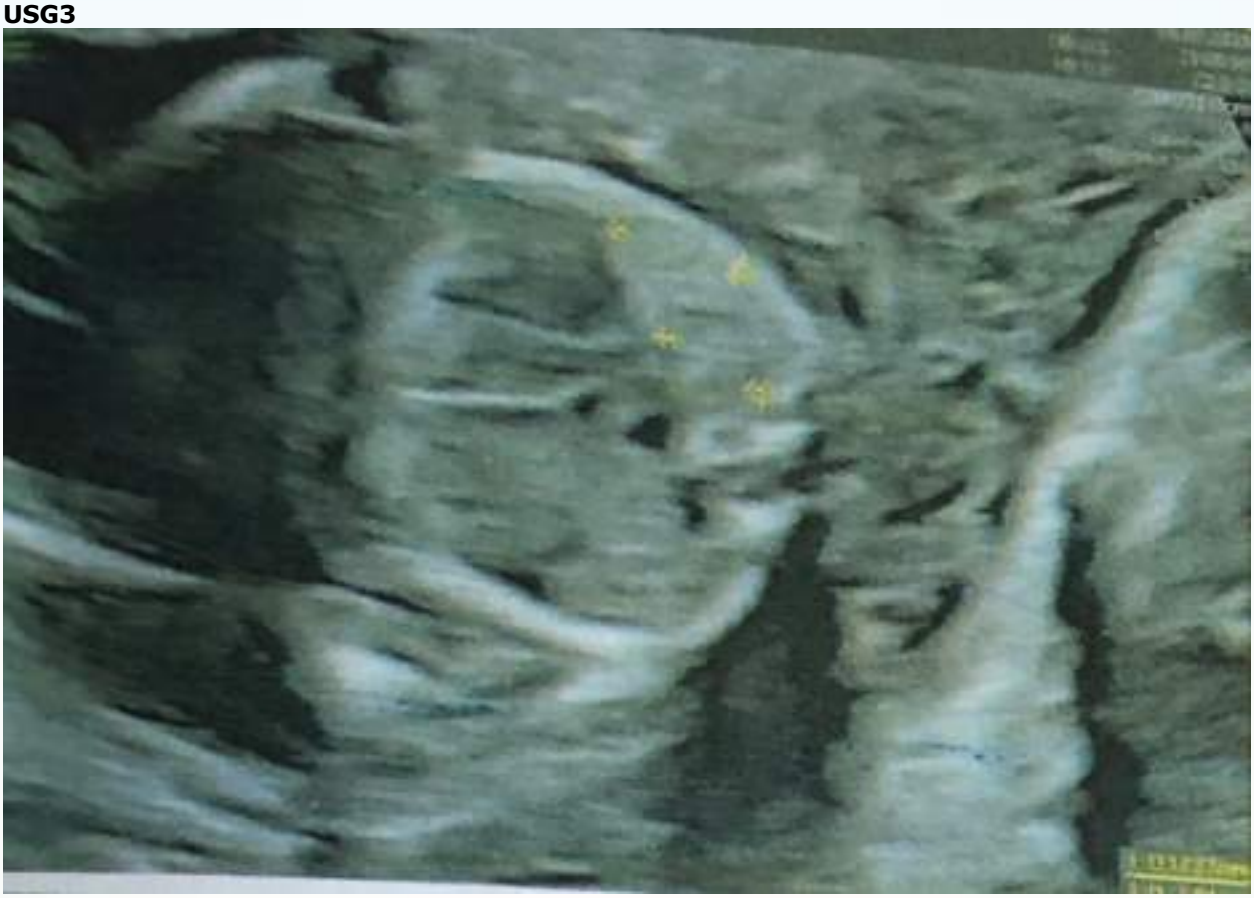
USG1


2nd Trimester / CA1-75 / 14.0cm / 32Hz
LMP 12-10-2024 / 18w2d / TIs 0.1 / TIb 0.1 / MI 1.0
[2D]
Res
Gn 47
DR 110
FA 4
P 90%

USG2


2nd Trimester / CA1-75 / 14.0cm / 32Hz
LMP 12-10-2024 / 18w2d / TIs 0.1 / TIb 0.1 / MI 1.0
[2D]
Res
Gn 47
DR 110
FA 4
P 90%

USG3



PS-036

İnfantil tip polikistik böbrek hastalığı

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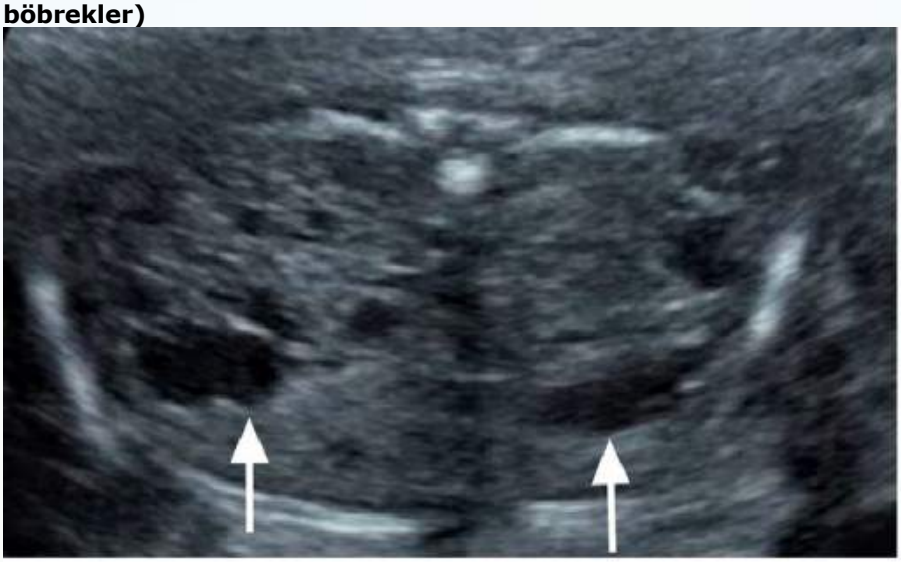
Bilateral kistik displastik böbrek ve otozomal resesif polikistik böbrek hastalığı (infantil tip) prenatal dönemde saptanan en ciddi üriner sistem anomalilerindendir. Bu anomalilerde, nefron gelişimindeki bozukluk ve duktal sistemdeki malformasyon sonucu böbreklerin normal yapısının yerini kistik, displastik dokunun almasıyla karakterizedir. Prenatal ultrasonografi sayesinde özellikle ikinci trimesterde tanı oranı belirgin şekilde artmıştır. Literatürde, bilateral olguların büyük çoğunluğunda oligohidramnios, pulmoner hipoplazi ve neonatal dönemde artmış mortalite riski bildirilmektedir. Çok merkezli çalışmalar, bu anomalilere sıklıkla başka santral sinir sistemi ve kardiyak bulguların eşlik edebildiğini ve genetik etiyolojinin heterojen olduğunu bildirmiştir.

OLGU: Otuz yedi yaşında G2.P1 olan hasta, 17. gebelik haftasında oligohidramnios nedeniyle yönlendirildi. Ayrıntılı ultrasonografide tüm batını dolduran, kortikomedüller ayrımı seçilemeyen, polikistik displastik görünümde bilateral genişlemiş böbrekler saptandı (Figür 1). Ek olarak bilateral milimetrik koroid pleksus kistleri ve üçüncü ventrikülde genişleme izlendi. Toplam amniyon sıvı indeksi 3 cm ölçülerek oligohidroamniyozis olarak değerlendirildi. Bu bulgularla olgu perinatoloji konseyinde infantil tip polikistik böbrek hastalığı ön tanısıyla değerlendirildi.Aileye olası genetik nedenler, neonatal mortalite, pulmoner hipoplazi ve uzun dönem prognoz hakkında kapsamlı bilgi verildi. Amniyosentez işleminş kabul etmedi. Ciddi fetal renal yetersizlik bulguları olan gebeye multidisipliner konsey kararı ve aile onamı ile 17+3 gebelik haftasında gebeliğin terminasyonu ailenin onamı alınarak uygulanmıştır (Figür 2). Bilateral infantil tip polikistik böbrek olgularında prognoz kötüdür. Ağır oligohidramnios pulmoner hipoplaziye yol açarak neonatal mortaliteyi artırır. Literatürde bu olguların çoğunda erken renal yetmezlik ve kısa yaşam bildirilmiştir. PAX2 ve HNF1B mutasyonları kistik displazi ve beyin anomalileriyle ilişkilidir. Güncel kılavuzlar ağır bilateral formlarda genetik test ve kapsamlı aile danışmanlığını önermektedir.

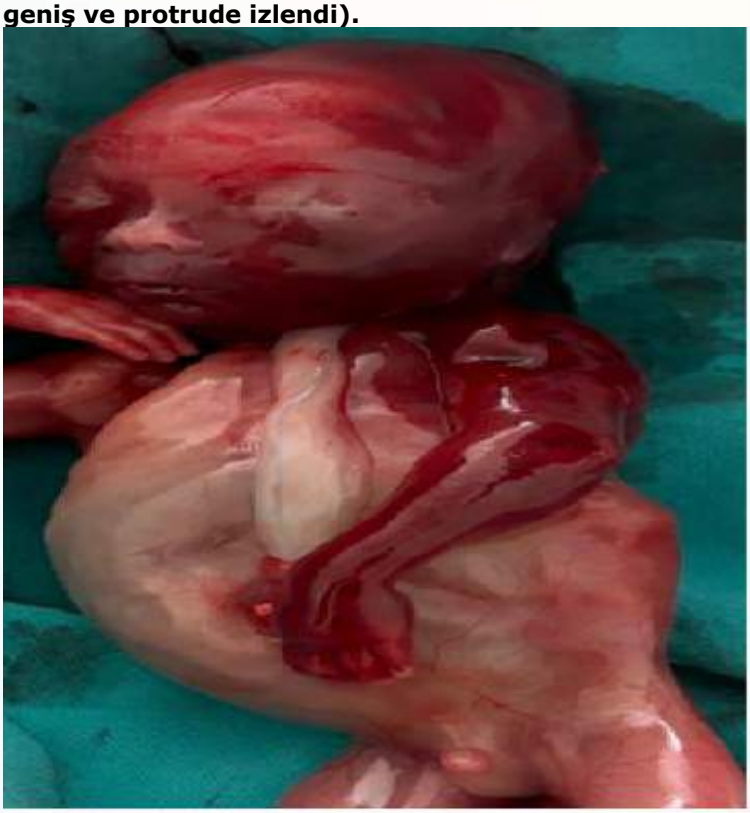
Anahtar Kelimeler: Renal anomalilar, Polikistik böbrek, Oligohidramnios

Figür 1: 17 hafta gebelikte oligo-anhidramnios ile birlikte bilateral infantil tip polikistik displastik böbrekler (ok işaretleri: fetal batının büyük kısmını dolduran ekspanse, displastik böbrekler)



17 hafta gebelikte oligo-anhidramnios ile birlikte bilateral infantil tip polikistik displastik böbrekler (ok işaretleri: fetal batının büyük kısmını dolduran ekspanse, displastik böbrekler)

Figür 2: Bilateral polikistik displastik böbrekleri olan olgunun postmortem görünümü (batın geniş ve protrude izlendi).



Bilateral polikistik displastik böbrekleri olan olgunun postmortem görünümü (batın geniş ve protrude izlendi).

PS-037

Dikoryonik Triamniotik Üçüz Gebeliklere Yaklaşım

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Dikoryonik triamniyotik (DCTA) üçüz gebelikler, özellikle yardımcı üreme teknikleri sonrası artış göstermekte olup maternal (preeklampsi, kardiyak hastalık), obstetrik (erken doğum, FGR) ve neonatal komplikasyonlar açısından yüksek risklidir. Yönetim seçenekleri arasında müdahalesiz izlem, monokoryonik çiftin veya ayrı plasentalı fetüsün redüksiyonu, tüm gebeliğin terminasyonu yer alır. Müdahalesiz izlemde ciddi prematürite oranı yüksektir. Fetal redüksiyon en uygun olarak 11–14. haftalarda, ense kalınlığı ölçümü ve erken anatomik tarama eşliğinde, intratorasik KCl enjeksiyonu ile gerçekleştirilir. 16. haftadan sonra kayıp ve erken doğum riski artarken, 32. hafta sonrası seçilmiş olgularda KCl veya lidokain ile güvenli fetosid yapılabilir. Monokoryonik çiftlerde vasküler anastomozlar nedeniyle klasik enjeksiyon teknikleri uygun değildir; radyofrekans ablasyon veya intrafetal lazer gibi yöntemler kullanılabilse de eş fetüs kaybı riski vardır. Literatür, üçüzden ikize indirgeme ile obstetrik komplikasyonlarda azalma olduğunu, ancak tekile indirgeme ile gebelik haftası ve neonatal sağkalımın daha avantajlı olduğunu göstermektedir.

Kendi serimizde 11–14. haftalarda MCDA ikizlerden ulaşılması kolay olan fetüse intratorasik KCl enjeksiyonu uygulandı. Müdahaleden sonraki kontrolde ikinci fetüste de kalp atımının durduğu izlendi ve gebelik tekile indirgenmiş oldu. Tüm olgularda (8/8) tekil gebelik elde edildi, invaziv işlem riski en aza indirildi. Sonuç olarak, DCTA gebeliklerde perinatoloji danışmanlığı esastır; komplikasyonları azaltma, doğum haftası ve doğum ağırlığını artırma hedefleri doğrultusunda tekile indirgeme en avantajlı strateji olarak öne çıkmaktadır.

Anahtar Kelimeler: gebelik, indirgeme, üçüz

DCTA Gebeliklerde Yönetim Tablosu-1

Tablo 1: Yönetim Seçenekleri ve Riskler

Yönetim Seçeneği	Avantajlar	Dezavantajlar / Riskler
Müdahalesiz izlem	Müdahale yok, doğal seyir	Ciddi prematürite riski yüksek
Üçüzden ikize redüksiyon	Obstetrik komplikasyonlarda azalma	Tekle indirgemeye göre daha erken doğum, daha düşük sağkalım
Üçüzden tekle redüksiyon	Doğum haftası daha geç, sağkalım daha iyi	Gebelik kaybı riski var, veri sınırlı
Monokoryonik çiftin bir fetüsünün redüksiyonu	[illegible]	Vasküler anastomozlar nedeniyle yüksek komplikasyon riski
Monokoryonik çiftin iki fetüsünün redüksiyonu	Tekil gebeliğe geçiş, en düşük prematürite	Düşük riski artabilir
Ayrı plasentalı fetüsün redüksiyonu	Monokoryonik çift korunur	En yüksek düşük oranı, daha erken doğum riski

DCTA Gebeliklerde Yönetim Tablosu-1

DCTA Gebeliklerde Yönetim Tablosu-2

Tablo 2: Redüksiyon Sonrası Gebelik Haftası ve Kayıp Oranları

Redüksiyon Sonrası Gebelik Tipi	Ortalama Gebelik Haftası	Gebelik Kaybı Oranı (%)
Tekil	38.0 hf	3.8
İkiz	35.2 hf	5.3
Üçüz	30.3 hf	-

DCTA Gebeliklerde Yönetim Tablosu-2

DCTA Gebeliklerde Yönetim Tablosu-3

Tablo 3: Literatür Bulguları (Standardize Edilmiş)

Çalışma	Popülasyon	Sonuç
Chaveeva et al., 2013	DCTA üçüz gebelikler	Redüksiyon ile preterm doğum azaldı
Morlando et al., 2025	Dikoryonik üçüz gebelikler	Redüksiyonda düşük riski ↑, ancak ciddi prematürite ↓
Hessami et al., 2022	DCTA üçüz gebeliklerde redüksiyon (meta-analiz)	Üçüz indirgeme ikize göre daha erken doğum ve daha düşük ağırlıklar
Meng et al., 2023	DCTA üçüz gebelikler	Müdahalesiz izlemde <34 hf doğum çok daha sık
Agarwal Kilis et al.	DCTA üçüz gebeliklerde vaka serisi (n=38)	Tekile indirgeme güvenli ve etkili

DCTA Gebeliklerde Yönetim Tablosu-3

PS-038

Prenatal Takiplerde Antenatal Tarama Testlerinin Trizomi 21 Tanısındaki Önemi: Olgu Serisi

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Olgu 1:36 yaşında, gebeliğinin 39.haftasında olan hasta su gelişi nedeniyle kliniğimize başvurdu.Gestasyonel Diyabet ve membran rüptürü tanılarıyla takip altına alındı.Antenatal dönemde düzenli takip edilmeyen ve hastaneye başvurmayan hastanın 1.trimester tarama testinde Trizomi 21 riskinin düşük risk olduğu görüldü.2.trimester ayrıntılı ultrasonografisi yapılmamıştı.24-28.hafta 75 gram OGTT sonucunda Gestasyonel Diyabet tanısı aldığı görüldü.39 hafta 2 günlük iken Fetal Distress nedeniyle acil sezaryen kararı alındı.

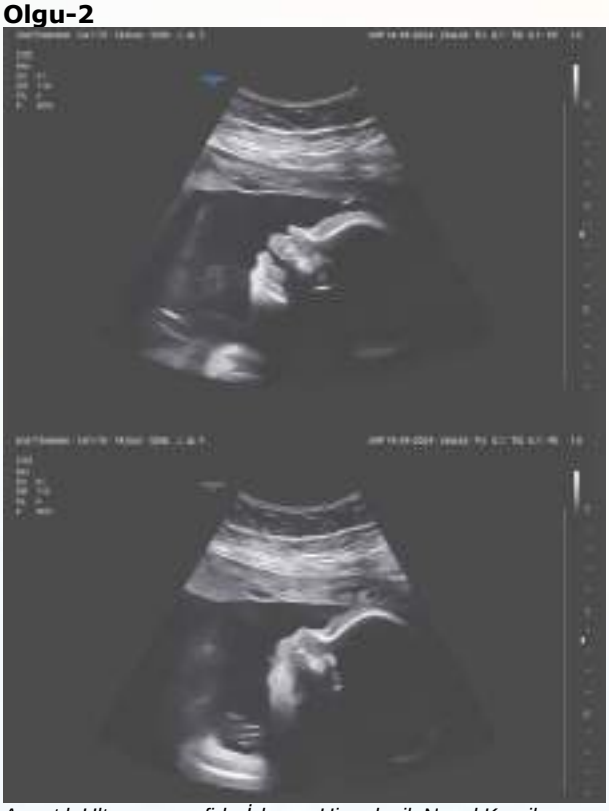
Apgar 1. Dakika 7, 5. Dakika 9 izlenen 2800 gram canlı kız bebek doğurtuldu.Postnatal muayenede çekik göz,burun kökü basıklığı,kısa parmaklar ve sandal gap deformitesi saptandı.Ekstremiteler arası satürasyon farkı pediatri ekibi tarafından kardiyak patoloji lehine değerlendirildi, simian çizgisi izlenmedi. Olgu 2:35 yaşında,gebeliğinin 20.haftasında ayrıntılı ultrasonografi amacıyla perinataloji polikliniğimize başvurdu.2.trimester tarama testinde Trizomi 21 için 1/56,Trizomi 18 için 1/206 saptadığımız hastanın ayrıntılı ultrasonografisinde küçük inlet ventriküler septal defekt ve nazal kemik hipoplazik izlendi.Trizomi 21 riski yüksek tespit edilen hastaya amniyosentez ve genetik danışmanlık önerildi.22 hafta 3 günlük iken amniyosentez yapılan hastanın QF-PCR ve uzun süreli hücre kültürü sonucu Trizomi 21 olarak raporlandı.Gebelik danışmanlık alan hasta gebeliğini sonlandırmaya karar verdi ve hastaya fetosit işlemi planlandı.Fetal ultrasonografi eşliğinde intrakardiyak 10 cc potasyum klorür(KCl) uygulanarak fetosit işlemi gerçekleşti. Misoprostol ile başlatılan indüksiyonun başarısız olması ve önceki sezaryen öyküsü nedeniyle histerotomi kararı alındı.Sezaryen ile 1095 gr, 36 cm ölçülerinde olan ex erkek bebek doğurtuldu.Postnatal muayenede basık burun, ense kalınlığı, simian çizgileri ve sol elde polidaktili saptandı.Prenatal dönemde yapılan tarama testleri ve gerek duyulduğunda invaziv tanı yöntemleri (amniyosentez, koryon villus örneklemesi) aileye doğru danışmanlık verilmesi açısından kritik öneme sahiptir.

Anahtar Kelimeler: Trizomi 21,Amniyosentez, Prenatal Ultrasonografi

Olgu-1

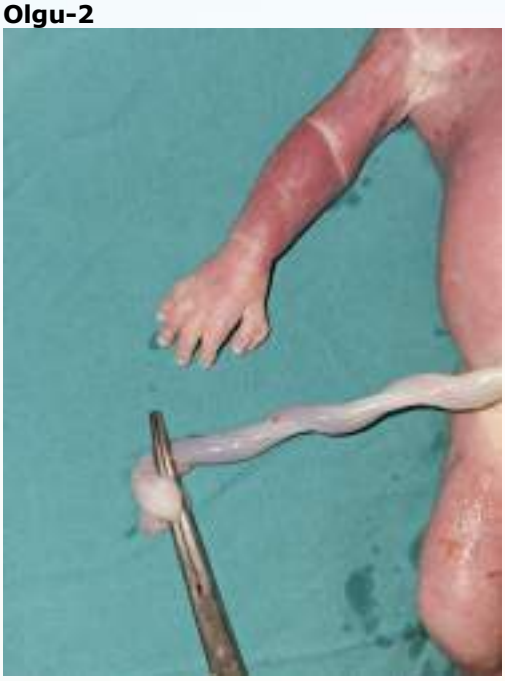
2. Trimester Ayrıntılı Ultrasonografisi Yapılmayan Anne Bebeği

Olgu-2



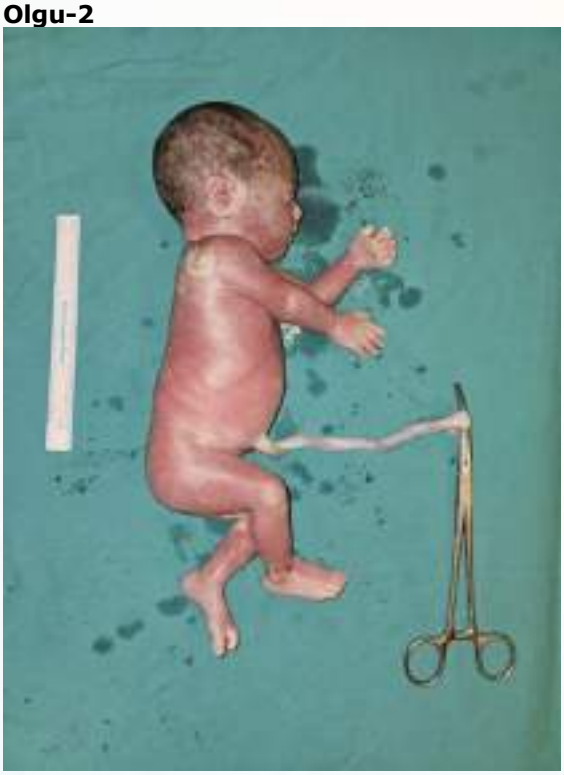
Ayrıntılı Ultrasonografide İzlenen Hipoplazik Nazal Kemik

Olgu-2



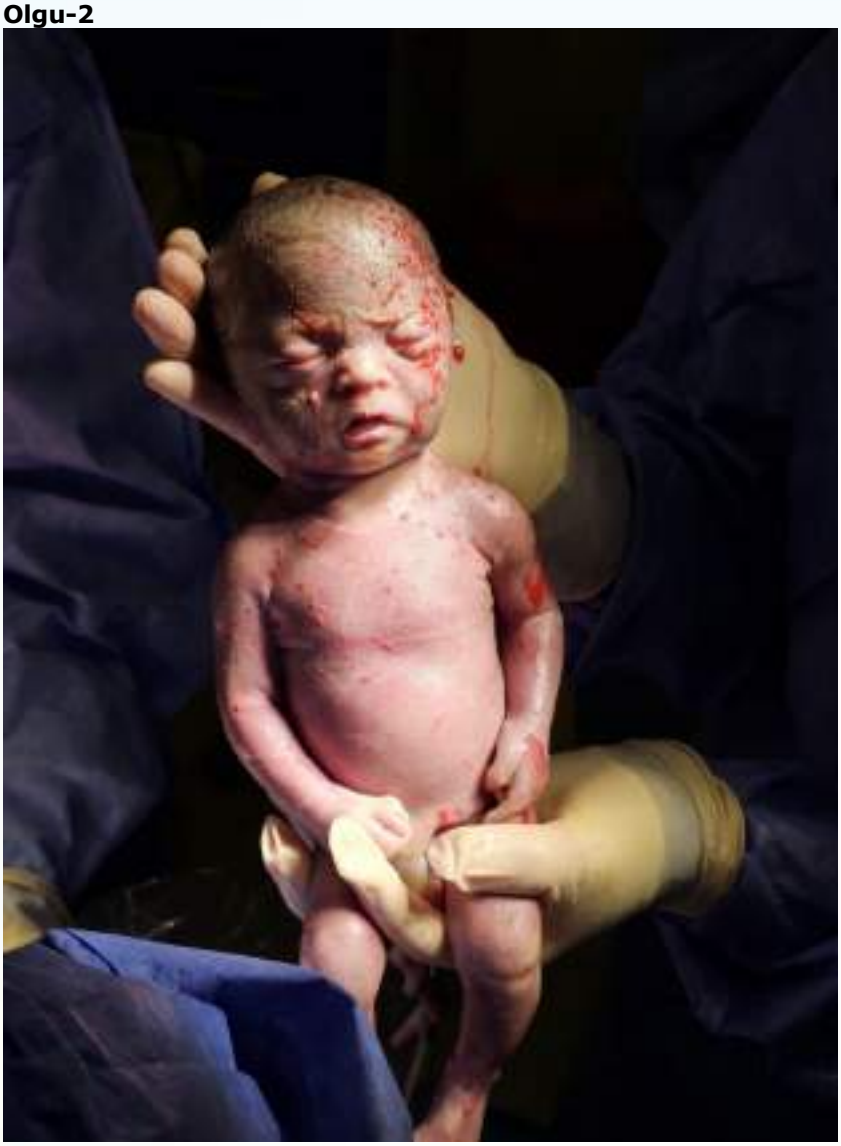
Fetosit Yapılan Trizomi 21 Bebekte Görülen Polidaktili

Olgu-2



Fetosit Yapılan Trizomi-21 Bebekte Görülen Simian Çizgisi Ve Ense Kalınlığı Bulguları

Olgu-2



Fetosit Yapılan Ex Erkek Bebek

PS-039

Inferior Vermian Hipoplazi ve Bilateral Displastik Böbrekler: Dismorfik Cavum Septum Pellucidum İle Kompleks Fetal Anomali

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GİRİŞ: Posterior fossa malformasyonları serebellum ve beyin sapını ilgilendiren, prenatal tanıda kritik öneme sahip anomalilerdir.Dandy–Walker malformasyonu,Blakes pouch kisti, mega sisterna magna ve izole vermian hipoplazi bu spektrumda yer alır.Görülme sıklığı yaklaşık 1/5000'dir. Prognoz,eşlik eden santral sinir sistemi veya sistemik anomalilere bağlıdır.Ultrasonografi temel tanı yöntemidir; fetal MRG ayırıcı tanıda önemlidir.Orta hat yapılarının incelenmesi de kritiktir.Özellikle cavum septum pellucidumda (CSP) uzunluk/en oranının 1,5'in altında olması "dismorfik CSP" olarak tanımlanır ve korpus kallozum agenezisi,septo-optik displazi veya kompleks malformasyonlarla ilişkili olup prognozu ağırlaştırabilir.

OLGU: 25 yaşında,G2P1 olan ve 26 hafta 2 günlük gebe, dış merkezde posterior fossa anomalisi saptanması üzerine tarafımıza refere edildi. Ayrıntılı ultrasonografide serebellar hemisferlerde ayrışma ve vermian agenezi izlendi.Posterior fossa anteroposterior çapı 15 mm ölçüldü (Figür1).CSP dismorfik izlenmiş olup uzunluk/eni oranı 0,84 olarak saptandı.Korpus kallozum doğal izlendi.Her iki böbrek belirgin şekilde büyümüş, hiperekojen ve multikistik görünümde izlenmiş, kortikomedüller ayrım kaybolmuştu (Figür2).Gebeye bu bulgularla Kordosentez uygulandı. Genetik inceleme FISH ve kültür sonuçları normal geldi.Perinatoloji konseyi değerlendirmesi sonrası ayrıntılı inceleme amacıyla fetal MRG önerildi.Yapılan MRG'de inferior vermian hipoplazi ve mega sisterna magna (CM:17,5 mm) saptandı.Her iki böbrek multikistik displastik olarak değerlendirildi.Tüm bulgularla olumsuz prognoz ve olası genetik sendrom riski nedeniyle gebeliğin sonlandırılması önerilen aileye genetik danışmanlık verildi ve gebeliğin 27+4 gebelik haftasında terminasyon kararı alındı.

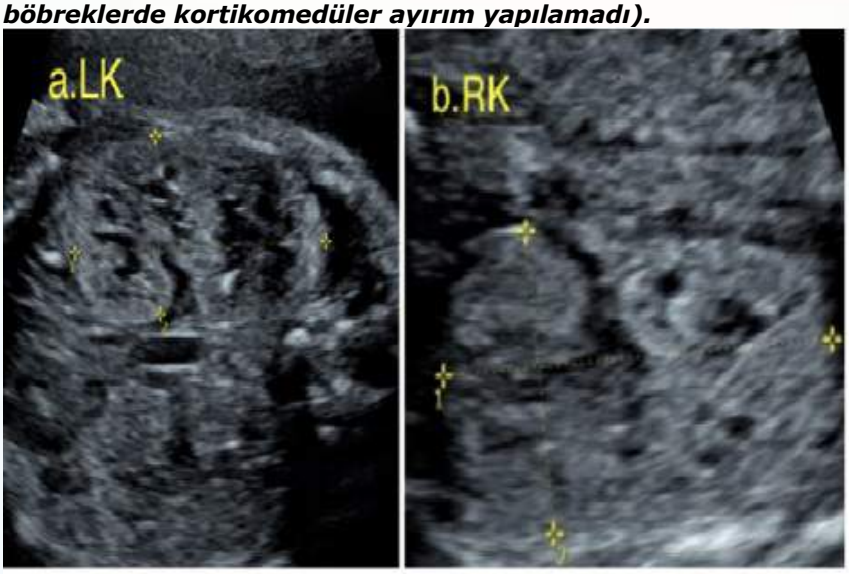
TARTIŞMA: Posterior fossa anomalilerinin yaklaşık yarısında ek anomali bildirilmiş,üçte birinde kromozomal bozukluk saptanmıştır.İki taraflı multikistik böbrek displazisi ise oligohidramniyos ve pulmoner hipoplazi nedeniyle neonatal mortaliteye yol açar. Serebellar vermis anomalileri ile böbrek displazisinin birlikteliği Joubert veya Meckel–Gruber sendromunu düşündürür.Bu vaka, kompleks fetal anomalilerde multidisipliner yaklaşım ve genetik danışmanlığın önemini göstermektedir.

Anahtar Kelimeler: Vermian hipoplazi, dismorfik CSP, Fetal displastik böbrekler

Figür 1. 26+2 hafta gebelikte dilate arka fossa, vermian hipoplazi, inferior serebellar ayrışma izlenen vakamızın 3.D ultrasonografisi. C: serebellum, V: vermis (vermiste anteroposterior mesafe 11,65mm ölçüldü), PF: dilate arka fossa (CM:15mm), ok iş

26+2 hafta gebelikte dilate arka fossa, vermian hipoplazi, inferior serebellar ayrışma izlenen vakamızın 3.D ultrasonografisi. C: serebellum, V: vermis (vermiste anteroposterior mesafe 11,65mm ölçüldü), PF: dilate arka fossa (CM:15mm), ok işareti elevasyona uğramış tentorium cerebelli.

Figür 2. 26+2 hafta gebelikte bilateral multikistik, ekspanse fetal böbrekler (LK: sol böbrek 42x23mm boyutlarında, RK:sağ böbrek 40x19,8mm boyutlarında ölçüldü, bilateral böbreklerde kortikomedüler ayırım yapılamadı).



26+2 hafta gebelikte bilateral multikistik, ekspanse fetal böbrekler (LK: sol böbrek 42x23mm boyutlarında, RK:sağ böbrek 40x19,8mm boyutlarında ölçüldü, bilateral böbreklerde kortikomedüler ayırım yapılamadı).

PS-040

Ağır Triventriküler Ventrikülomegali ile Birlikte Parsiyel Rhombensefalosynapsis Olgusu

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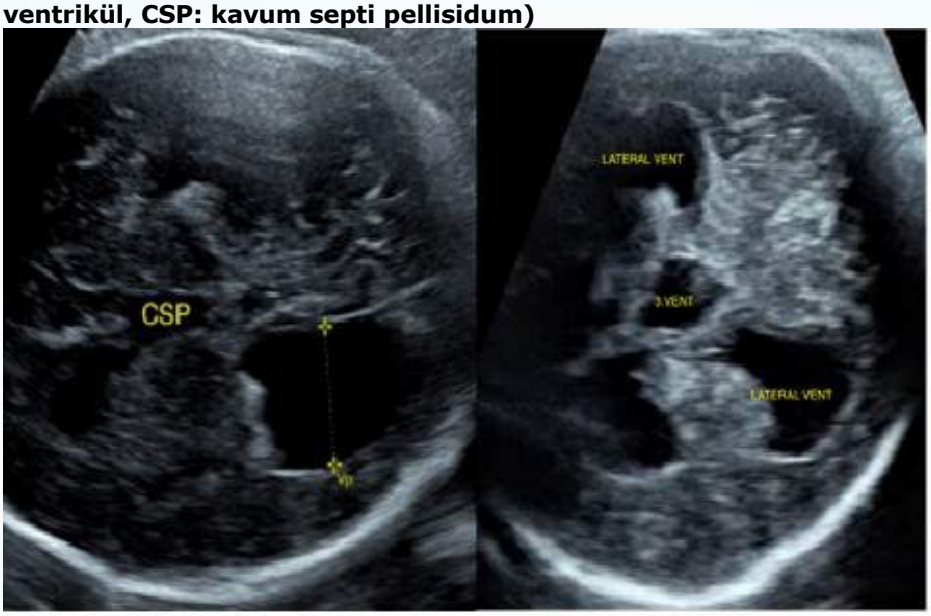
Rhombensefalosynapsis,serebellar vermisin kısmi ya da tam yokluğu ve serebellar hemisferlerin orta hatta birleşmesiyle karakterize nadir posterior fossa malformasyonudur. Embriyonik dönemde 5–6. haftalarda gelişimsel bozukluk sonucu ortaya çıkar.Sıklığı milyonda 1'den azdır.Prenatal tanıda en sık bulgu posterior fossada anormal serebellar morfoloji ve ventrikülomegalidir.Özellikle üçüncü ventrikül genişlemesi ile birlikte triventriküler ağır ventrikülomegalide prognoz olumsuzdur.25 yaşında,G3P2,34 + 5 haftalık gebe tarafımıza kraniyal yapıların ayrıntılı değerlendirilmesi amacıyla yönlendirildi.Kliniğimizde yapılan ayrıntılı ultrason incelemede sol lateral ventrikül çapı 23 mm,sağ lateral ventrikül çapı 17 mm ölçüldü,üçüncü ventrikül ise geniş bulunarak ağır triventriküler ventrikülomegali tanısı konuldu (Figür 1).Serebellar hipoplazi mevcuttu ve vermis izlenmedi.Vakaya mega cisterna magna eşlik etmekteydi(CM:13mm).Ek olarak böbreklerin kortikomedüller ayrımı anormal olmakla birlikte grade 2,3 hidronefroz ve sağ üreterde dilatasyon izlendi.Mevcut bulgular parsiyel rhombensefalosynapsis olasılığını düşündürdü (Figür 2).Fetal MR incelemesinde asimetrik ağır ventrikülomegali izlendi.Perinatoloji konseyinde olgu triventriküler ağır ventrikülomegali ve parsiyel rhombensefalosynapsis tanılarıyla tartışıldı, aileye olası genetik sendromlar,nörogelişimsel prognoz ve postnatal takip gereksinimleri hakkında ayrıntılı danışmanlık verildi.Gebelik terminasyonu ve invaziv işlemi kabul etmeyen hasta 39. gebelik haftasında sezaryen ile doğurtuldu.1.ve 5.dk APGAR skoru 8 ve 9 olan 2860 gram ölçülerinde kız bebek doğurtuldu.Yenidoğanın nörolojik değerlendirmesi doğum anında normal olarak değerlendirildi.Transfontanel USG'de solda belirgin olmak üzere her iki lateral ventrikül dilate görünümde, 3.ventrikül hafif belirgin izlendi.Vermis net izlenememiş olup 4.ventrikül seçilememiştir.RES çoğunlukla ventrikülomegali ile birliktedir ve prognoz eşlik eden anomalilerle belirlenir.İzole ağır ventrikülomegalide dahi nörogelişimsel sorun oranının %50'yi aştığını göstermektedir.Üçüncü ventrikül genişlemesi eklenince mortalite ve morbidite artar. RES, VLDLR ve RELN mutasyonlarıyla ilişkilidir; prenatal genetik test önerilir. Bu olgu, prenatal tanıda ultrason ve MR'ın tamamlayıcı rolünü göstermektedir.

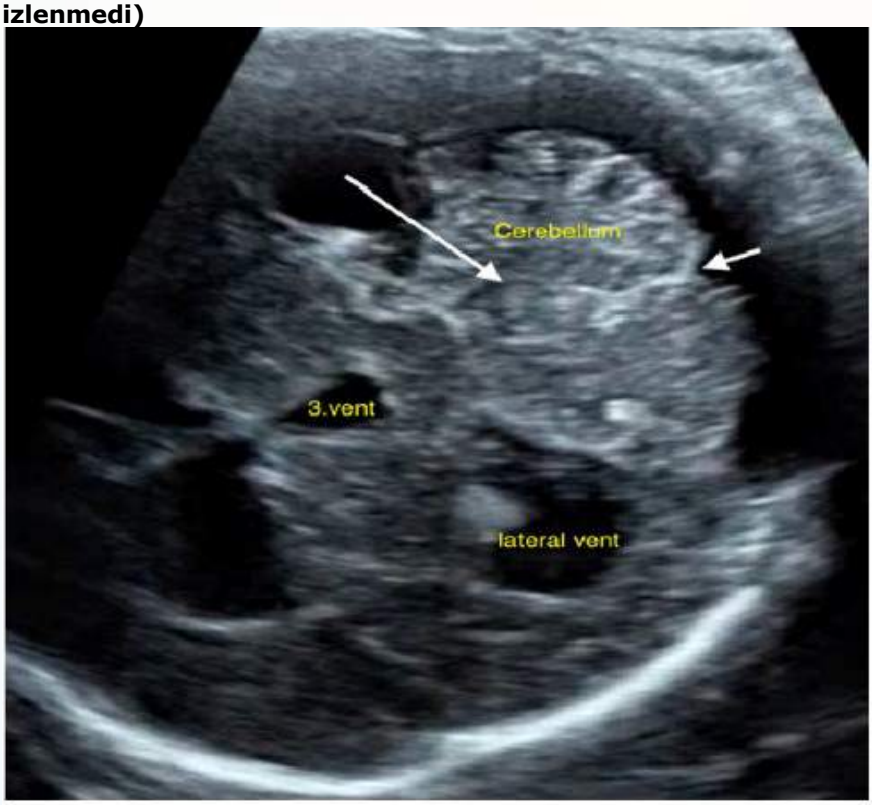
Anahtar Kelimeler: Rombansefalosinapsis, Ağır triventrikülomegali, Dilate üçüncü ventrikül

Figür 1: 34+5 hafta gebede ağır triventriküler ventrikülomegali(3. ventrikül, lateral ventrikül, CSP: kavum septi pellisidum)



34+5 hafta gebede ağır triventriküler ventrikülomegali(3. ventrikül, lateral ventrikül, CSP: kavum septi pellisidum)

Figür 2: 34+5 hafta gebede triventriküler ventrikülomegali, serebellar hipoplazi ve parsiyel rhombensefalosinapsis(kısa ok işareti: vermis izlenmedi, uzun ok işareti: 4. ventrikül izlenmedi)



34+5 hafta gebede triventriküler ventrikülomegali, serebellar hipoplazi ve parsiyel rhombensefalosinapsis(kısa ok işareti: vermis izlenmedi, uzun ok işareti: 4. ventrikül izlenmedi)

PS-041

Prenatal Tanıda Dandy–Walker Malformasyonu: Detaylı Ultrason Bulguları ve Yönetim

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Dandy–Walker malformasyonu (DWM), arka fossa gelişim bozuklukları spektrumunun önemli bir tipidir. Dördüncü ventrikülün kistik dilatasyonu, serebellar vermisin agenezi/hipoplazisi ve posterior fossanın genişlemesi ile karakterizedir. Prenatal tanı konulan olguların yaklaşık üçte birinde ek santral sinir sistemi, üçte birinde ise kardiyak, renal veya gastrointestinal anomaliler eşlik eder. Erken tanı; aileye prognoz, nörolojik sekel riski, genetik sendrom olasılığı ve gebelik yönetimi açısından kritik öneme sahiptir. DWM'ye sıklıkla trizomi 18,13 ve mikrodelesyon/duplikasyon sendromları eşlik eder. Bu nedenle ayrıntılı ultrasonografi ve genetik inceleme prenatal yönetimin temelini oluşturur.

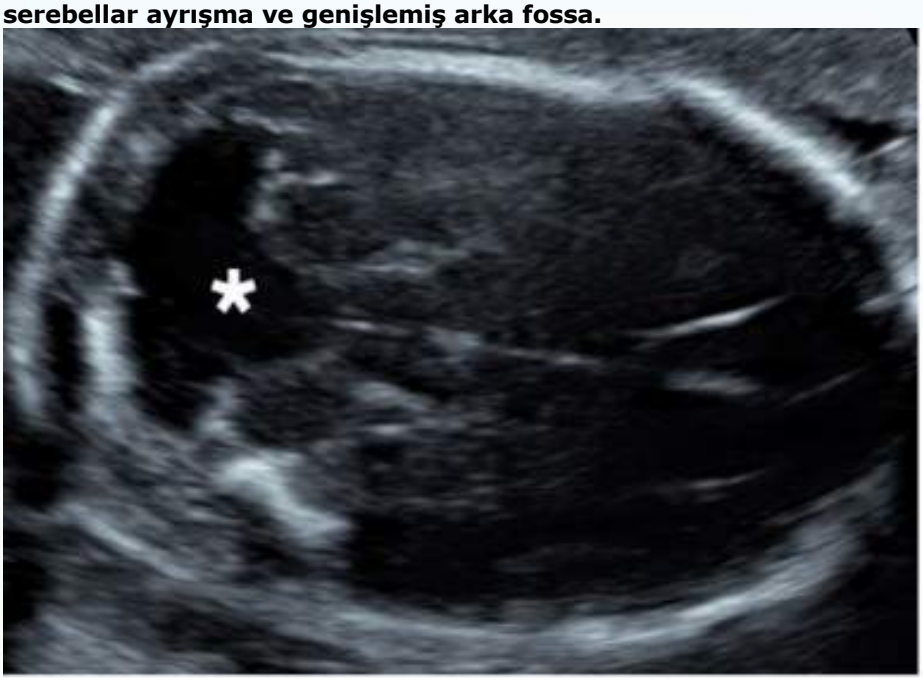
OLGU: 28 yaşında, G1P 0 olan hasta 19 + 4 haftalık gebeliği sırasında dış merkezde posterior fossa anomalisi şüphesiyle kliniğimize refere edildi. Ayrıntılı ultrasonografide posterior fossa belirgin şekilde genişlemiş, serebellar hemisferler ayrışık ve serebellar vermisin parsiyel agenezisi izlendi (Figür 1ve2). Nuchal kalınlık 6 mm ölçüldü, üçüncü ventrikül dilate izlendi. Ponto-vermian açının %29 oranında artmış olduğu belirlendi (Figür2). Ayrıca sağ lateral ventrikülde 8 × 7 mm boyutlarında koroid pleksus kisti ile grade 1 hiperekojen bağırsak dikkati çekti. Bu bulgular Dandy–Walker malformasyonu ve olası kromozomal anomali şüphesini kuvvetlendirdi.

Perinatoloji konseyinde değerlendirildi. Gebeye aynı haftada amniosentez uygulandı. Ayrıntılı danışmanlıkta olası nörogelişimsel sekeller, neonatal mortalite riski ve eşlik edebilecek diğer organ anomalileri anlatıldı. Konsey değerlendirmesi ve aile onamı sonrasında, Dandy–Walker malformasyonu ön tanısıyla gebeliğin sonlandırılmasına karar verildi ve terminasyon işlemi 20+4 gebelik haftasında gerçekleştirildi. Amniyosentez sonucunda kromozomal anomali saptanmadı.

DWM, prenatal dönemde tanınabilen en önemli posterior fossa anomalilerindendir. Prognoz, ek anomaliler ve genetik bulgularla yakından ilişkilidir. İzole olgularda bile %20 kromozom anomalisi bildirilmiştir. Yönetimde detaylı ultrason, kromozom analizi ve genetik danışmanlık esastır.

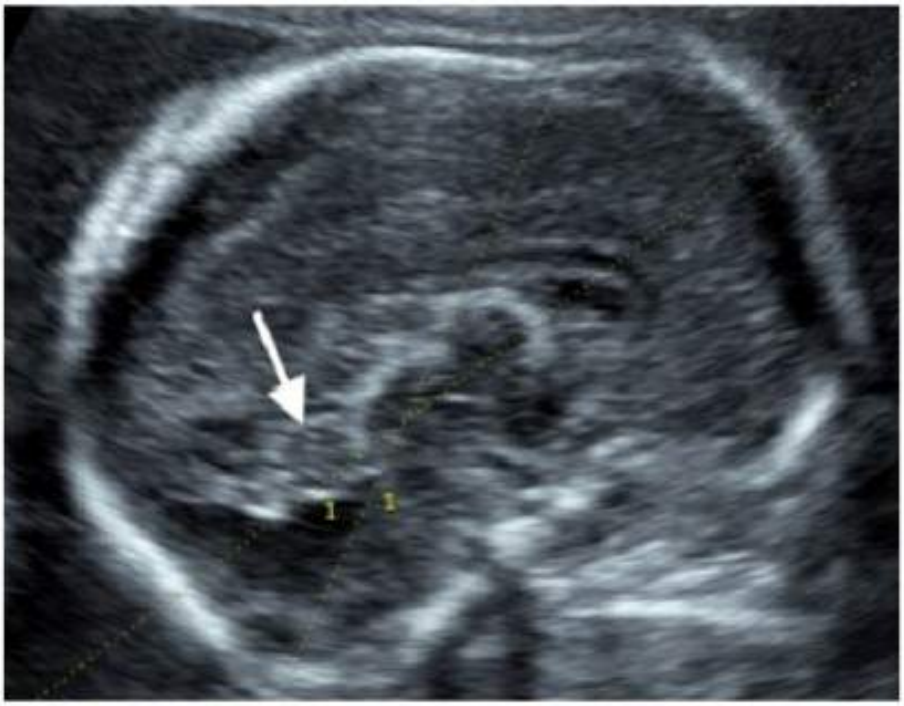
Anahtar Kelimeler: Arka fossa anomalisi, dandy- walker malformasyonu, prenatal tanı

Figür 1: 19+4 hafta gebelikte tanı alan Dandy Walker malformasyonu. Yıldız işareti: serebellar ayrışma ve genişlemiş arka fossa.



19+4 hafta gebelikte tanı alan Dandy Walker malformasyonu. Yıldız işareti: serebellar ayrışma ve genişlemiş arka fossa.

Figür 2: Artmıs pontovermian açı ve parsiyel vermian agenezi (ok işareti)



Artmıs pontovermian açı ve parsiyel vermian agenezi (ok işareti)

PS-042

Multipl Büyük Plasental Kistlerle Birlikte Bulunan Fetal Büyüme Geriliği ve Preeklampsi Olgusu

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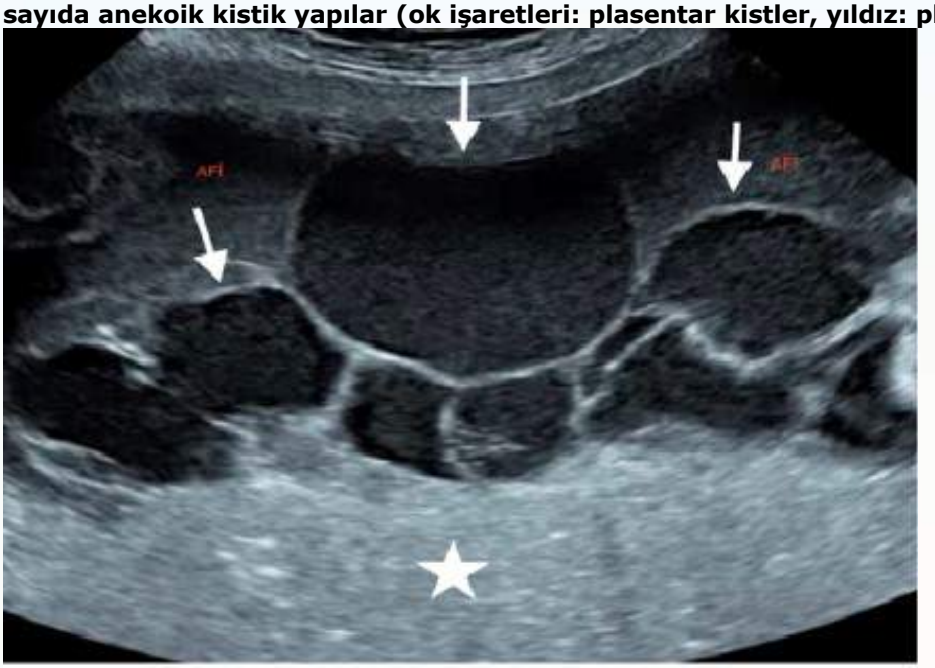
Plasental kistler, prenatal ultrasonografide tesadüfen saptanan, iyi sınırlı anekoik lezyonlardır.Tek ve küçük kistler genellikle klinik açıdan önemsizdir;ancak çok sayıda veya ≥4,5–5 cm boyutunda olduklarında uteroplasental perfüzyonu bozarak fetal büyüme geriliği (FGR), preeklampsi ve preterm doğum riskini artırabilirler.Literatürde bu olguların yakın ultrason-Doppler takibi ile doğum sonrası histopatolojik incelemenin önemi vurgulanmaktadır.29 yaşında,G7P3A3, 28 + 2 gebelik haftasında yapılan ultrasonografide posterior yerleşimli plasentanın anterior yüzünde,en büyüğü 39 × 39 mm ölçülerinde çok sayıda saydam,anekoik,multikistik yapılar izlenirken (Figür1ve2); female fetusta eşlik eden majör yapısal anomali saptanmadı.Perinatoloji konseyinde olgu "plasental kist" lehine değerlendirildi, doğum sonrası plasentanın patolojik ve genetik incelemesi planlandı.Hasta invaziv prenatal tanı testlerini kabul etmedi ve seri büyüme,Doppler takibi önerildi.32+2 gebelik haftasında yapılan kontrolde beklenen doğum ağırlığı 3 persantilin altında izlendi.Plasenta anterior yüzünde yaklaşık 5 cm boyutlarında çok sayıda kist görüldü ve aynı haftada yeni başlangıçlı preeklampsi gelişmesi üzerine gebeye yatış önerildi. Hastaya betametazon ve nöröprotektif amaçlı MgSO4 tedavisi uygulandı. 32+6 haftada yapılan NST-de tekrarlayan geç deselerasyonlar izlenmesi üzerine FGR,ağır preeklampsi ve NST'deki patolojik bulgular nedeniyle acil sezaryen ile doğum gerçekleştirildi, 1500 gr, 43 cm ölçülerinde 1.ve 5. dk APGAR sokuru 7 ve 9 olan kız bebek doğurtuldu.Plasentanın fetal yüzünde bir miktar makroskopik kistik yapılar vizüalize edildi (Figür3). Bu olgu, büyük ve çoklu plasental kistlerin FGR'ye ek olarak erken preeklampsi ve fetal distres ile ilişkili olabileceğini göstermektedir.Literatür, 5 cm'den büyük/multipl kistlerde perinatal komplikasyon riskinin arttığını bildirmektedir. Yönetimde seri biyometri, umbilikal arter,MCA,duktus venozus Dopplerleri, sık fetal iyilik testleri ve antenatal steroid uygulaması önerilir. Maternal/fetal durumda bozulma geliştiğinde doğum geciktirilmemelidir.

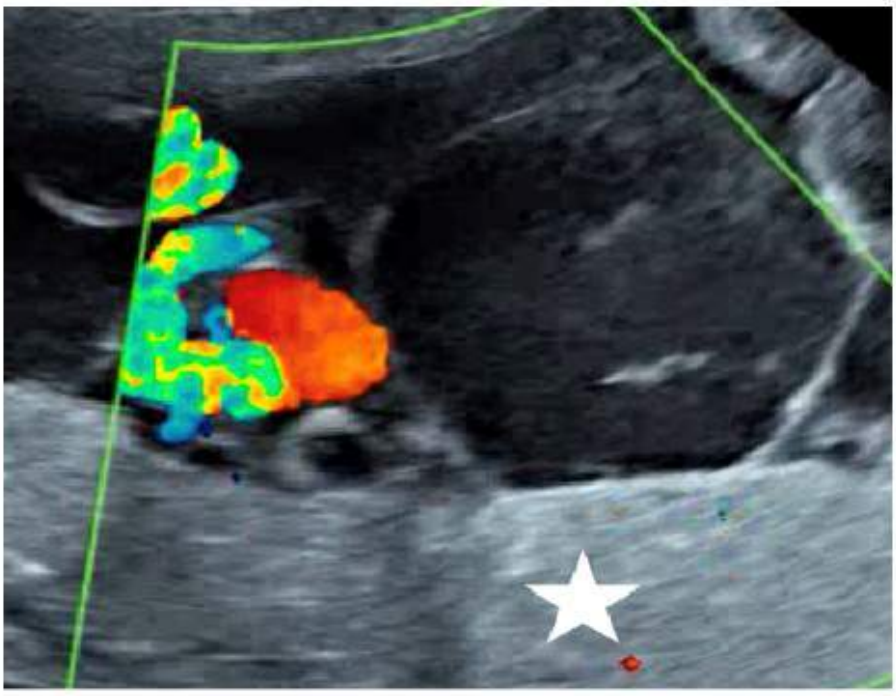
Anahtar Kelimeler: Plasentar kist, Fetal gelişme geriliği, Preeklampsi

Figür 1: 28+2 hafta gebelikte plasentanın fetal yüzünde büyüyü 4 cm boyutlarında çok sayıda anekoik kistik yapılar (ok işaretleri: plasentar kistler, yıldız: plasenta)



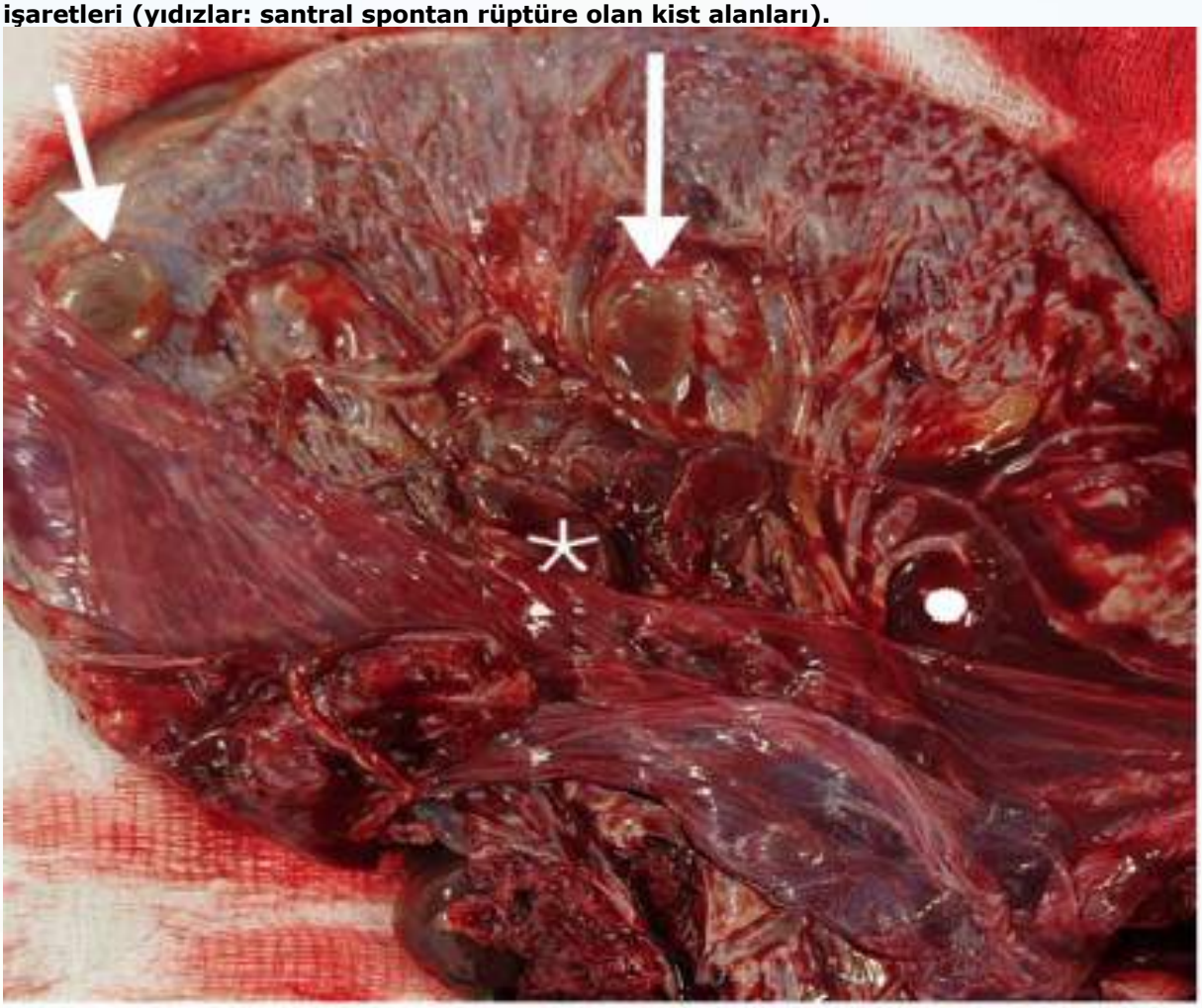
28+2 hafta gebelikte plasentanın fetal yüzünde büyüyü 4 cm boyutlarında çok sayıda anekoik kistik yapılar (ok işaretleri: plasentar kistler, yıldız: plasenta)

Figür 2: Plasentar kistlerin umblikal kord ile ilişkisi: umblikal kordun plasentaya insersiyon alanın önünde vasküler aktif olmayan çok sayıda anekoik kistik yapılar (yıldız: plasenta).



Plasentar kistlerin umblikal kord ile ilişkisi: umblikal kordun plasentaya insersiyon alanın önünde vasküler aktif olmayan çok sayıda anekoik kistik yapılar (yıldız: plasenta).

Figür 3: Sezaryen ile doğum, plasentar kistlerin spontan rüptüre olmamış bir kısmı- ok işaretleri (yıdızlar: santral spontan rüptüre olan kist alanları).



Sezaryen ile doğum, plasentar kistlerin spontan rüptüre olmamış bir kısmı- ok işaretleri (yıdızlar: santral spontan rüptüre olan kist alanları).

PS-043

Birinci Trimesterde Tanı Konulan Alobar Holoprosensefali İle Karakterize Triploidi Vakası

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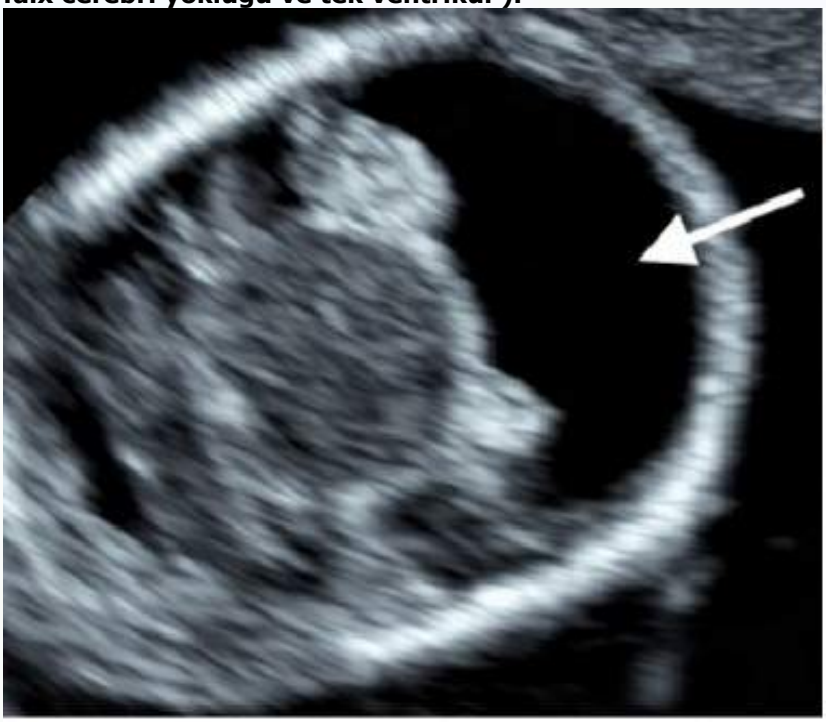
Holoprosensefali (HPE),ön beyin veziküllerinin embriyonik dönemde tam olarak ayrılmamasıyla karakterize,insidansı canlı doğumlarda yaklaşık 1:10.000–20.000 olan ağır bir santral sinir sistemi malformasyonudur.Alobar tip,serebral hemisferlerin ve ventriküllerin tamamen birleşik olduğu en ağır formdur ve çoğunlukla letaldir.HPE olgularının %30–50'sinde kromozomal anomaliler eşlik eder;en sık trizomi 13 olmakla birlikte triploidi,trizomi 18 ve çeşitli mikrodelesyonlar da tanımlanmıştır.Prenatal ultrasonografi,özellikle ikinci trimesterde,alobar holoprosensefali tanısında temel görüntüleme yöntemidir.Orta hat yapılarının(falx cerebri,koroid pleksus ayrışması) izlenememesi ve tek ventrikül görünümü tipiktir.Genetik inceleme ise prognoz ve aile danışmanlığı açısından kritik öneme sahiptir. 33 yaşında G2P1,11+6 gebelik haftasında değerlendirilmiştir.Prenatal ultrasonografide beyin orta hattında falx yapısı izlenmemiş ve tek ventrikül yapısıyla uyumlu alobar holoprosensefali tespit edilmiştir(Figür1).Ek olarak fetal ellerde kapalı duruş(clenched hand) saptanmıştır (Figür2).Bulgular Perinatoloji Konseyi'nde değerlendirildi.CVS işlemini kabul etmeyen hastada gebelik,ailenin onayı ile sonlandırılmıştır(Figür3).Abort materyalinden genetik incelemede triploidi saptanmıştır. Alobar holoprosensefali letal,ciddi nörolojik ve yüz orta hat defektleriyle ilişkili anomalidir.Prenatal tanıda en kritik bulgu,11–13. gebelik haftalarından itibaren orta hattın izlenmemesi ve tek ventrikül görünümüdür.Olgumuzda ek olarak "clenched hand" bulgusu,triploidi veya trizomi 18 gibi anöploidileri akla getirmiştir.Triploidi,çoğu kez paternal dizomik ya da maternal monozomik gamet hatalarına bağlı gelişir ve insidansı 1:3.500 gebelik civarındadır;canlı doğum nadirdir.Holoprosensefali triploidi birlikteliği, letal seyre katkıda bulunur ve aileye prognoz hakkında kesin bilgi vererek erken terminasyon kararını kolaylaştırır.

Bu olgu,birinci trimesterde dahi alobar holoprosensefali ve ek ekstremite bulgularının dikkatli taramayla saptanabileceğini göstermektedir. CVS kabul etmeyen ailelerde,abort materyalinden genetik analiz yapılması,etiyolojiyi aydınlatmak ve rekürrens riskini değerlendirmek açısından önemlidir.Tekrarlama riski genellikle düşüktür,ancak ileri maternal yaş ve gamet dizomisi riski nedeniyle sonraki gebeliklerde ilk trimester kombine test ve erken hedef ultrason önerilir.

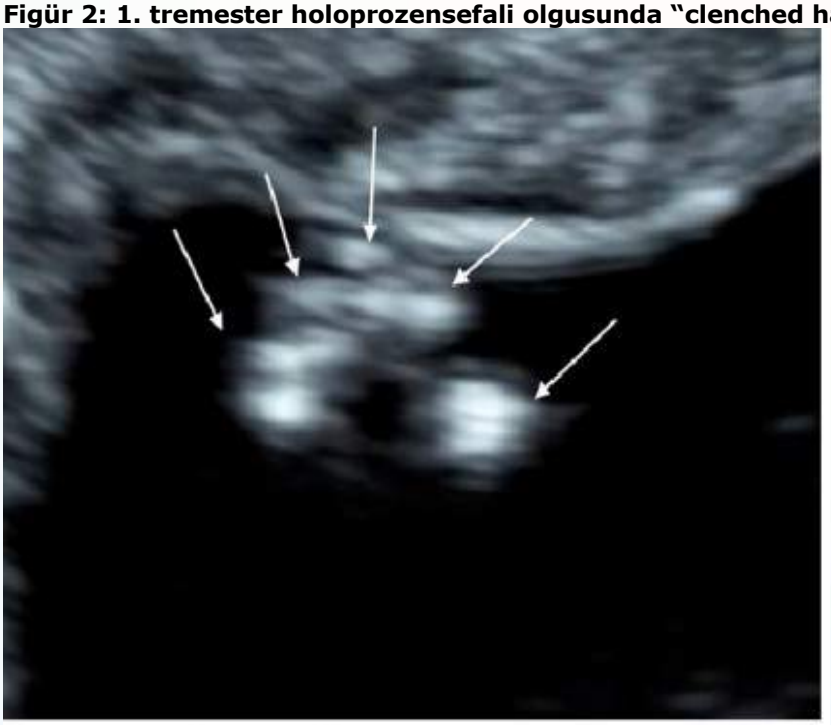
Anahtar Kelimeler: Alobar holoprozensefali, Triploidi, Clenched hand

Figür 1: Alobar holoprozensefali, füzyone talamuslar, tek ventrikül görünümü (ok işareti: falx cerebri yokluğu ve tek ventrikül).



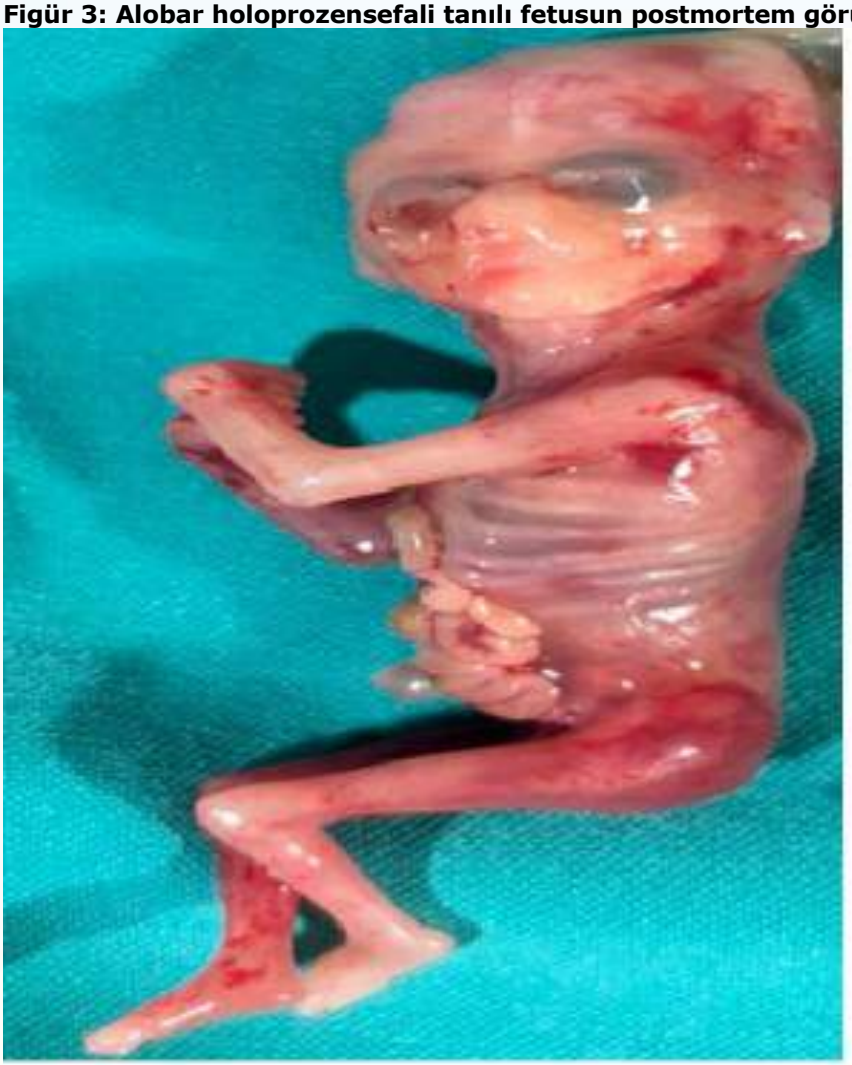
Alobar holoprozensefali, füzyone talamuslar, tek ventrikül görünümü (ok işareti: falx cerebri yokluğu ve tek ventrikül).

Figür 2: 1. tremester holoprozensefali olgusunda "clenched hand" görünümü.



1. tremester holoprozensefali olgusunda "clenched hand" görünümü.

Figür 3: Alobar holoprozensefali tanılı fetusun postmortem görünümü.



Alobar holoprozensefali tanılı fetusun postmortem görünümü.

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