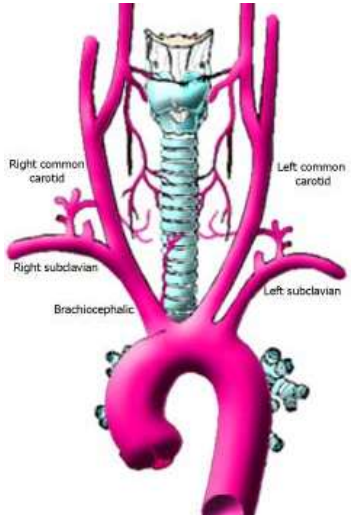
İZOLE VE İZOLE OLMAYAN ARSA OLGULARINDA TRİZOMİ 21 PREVALANSI

Dr. Aytül Çorbacıoğlu Esmer

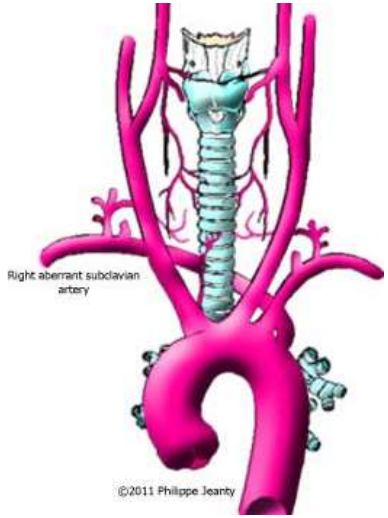
İstanbul Üniversitesi İstanbul Tıp Fakültesi
Perinatoloji Bilim Dalı

Aberan sağ subklavian arter

- En sık aortik ark anomalisi (otopsi serilerinde %1.2)
- Brakiosefalik arter yerine arkus aortadan sol subklavian arterin distalinden
- Trakea ve ösefagusun arasından geçerek sağ kola ilerler
- Genellikle asemptomatik, nadiren disfaji ve dispne



Sağ subklavian arter



Aberan sağ subklavian arter

Aberan sağ subklavian arter

- Fetuslarda kromozom anomalilerle ilişkili (özellikle T 21)
  - Trizomi 21 olgularında insidansı: %7.8-37.5
  - Kromozomu normal fetuslarda insidansı:%0.6-1.5

(Willruth, Ultraschall Med 2012, Borenstein Ultrasound Obstet Gynecol 2010, Zalel, Ultrasound Obstet Gynecol 2008, Borenstein Ultrasound Obstet Gynecol 2008)

  - Pozitif LHR: 21.48
  - Negatif LHR: 0.71

(Agathokleus, Ultrasound Obstet Gynecol 2013)

- İzole marker? Diğer bulgularla beraber anlamlı?

Aberan sağ subklavian arter

- İzole ARSA ve T 21 ilişkisi çelişkili
- LHR: 3.94 (Agathokleus, Ultrasound Obstet Gynecol 2013)
- Pozitif ve negatif LHR: 0 ve 1.005 (Yazıcıoğlu, Prenat Diagn 2013)

ARSA + T 21 olgularında izole ARSA insidansı

	Isole ARSA	Toplam ARSA	%
Chaoui et al. (2005)[1]	1	5	20
Zalel et al. (2008)[5]	0	3	0
Borenstein et al (2010)[4]	1	12	8.3
Rembouskos et al.(2010)[8]	1	6	16.6
Paladini et al. (2012)[9]	8	27	29.6
Willruth et al. (2012)[3]	0	1	0
Yazicioglu et al. (2013)[10]	0	7	0

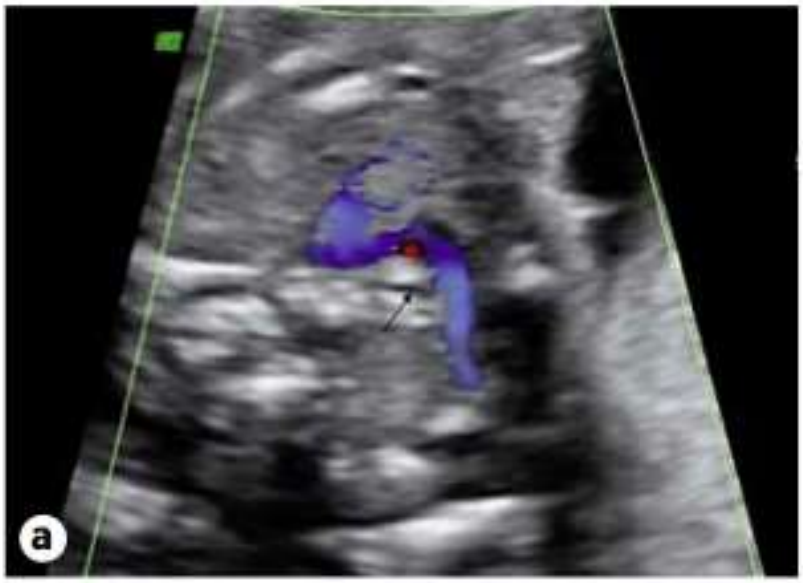
Amaç

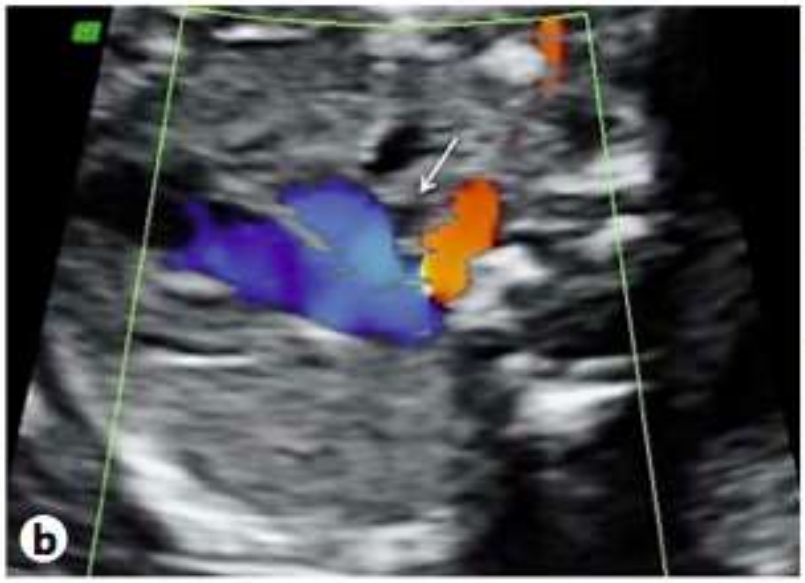
- İzole ve izole olmayan prenatal ARSA olgularında Down sendromu sıklığını ortaya koymak
- ARSA ile birlikteliği olan ultrason bulgularını incelemek

Gereç ve Yöntemler

- Retrospektif
- Ocak 2008-Aralık 2012
- Üç merkez
  - İstanbul Tıp Fakültesi
  - Kanuni Sultan Süleyman Eğitim ve Araştırma Hastanesi
  - İstanbul Prenatal
- Transabdominal USG
  - Voluson 730 Expert, GE Healtcare
  - Xario SSA-66A, Toshiba Medical Systems

Gereç ve Yöntemler





Gereç ve Yöntemler

- Hem düşük hem yüksek riskli hastalar
- Detaylı USG ve EKO
- İnvaziv prenatal (AS/KS) veya postnatal karyotip analizi
- FM normal olan yenidoğanın karyotipi normal olarak kabul edildi.

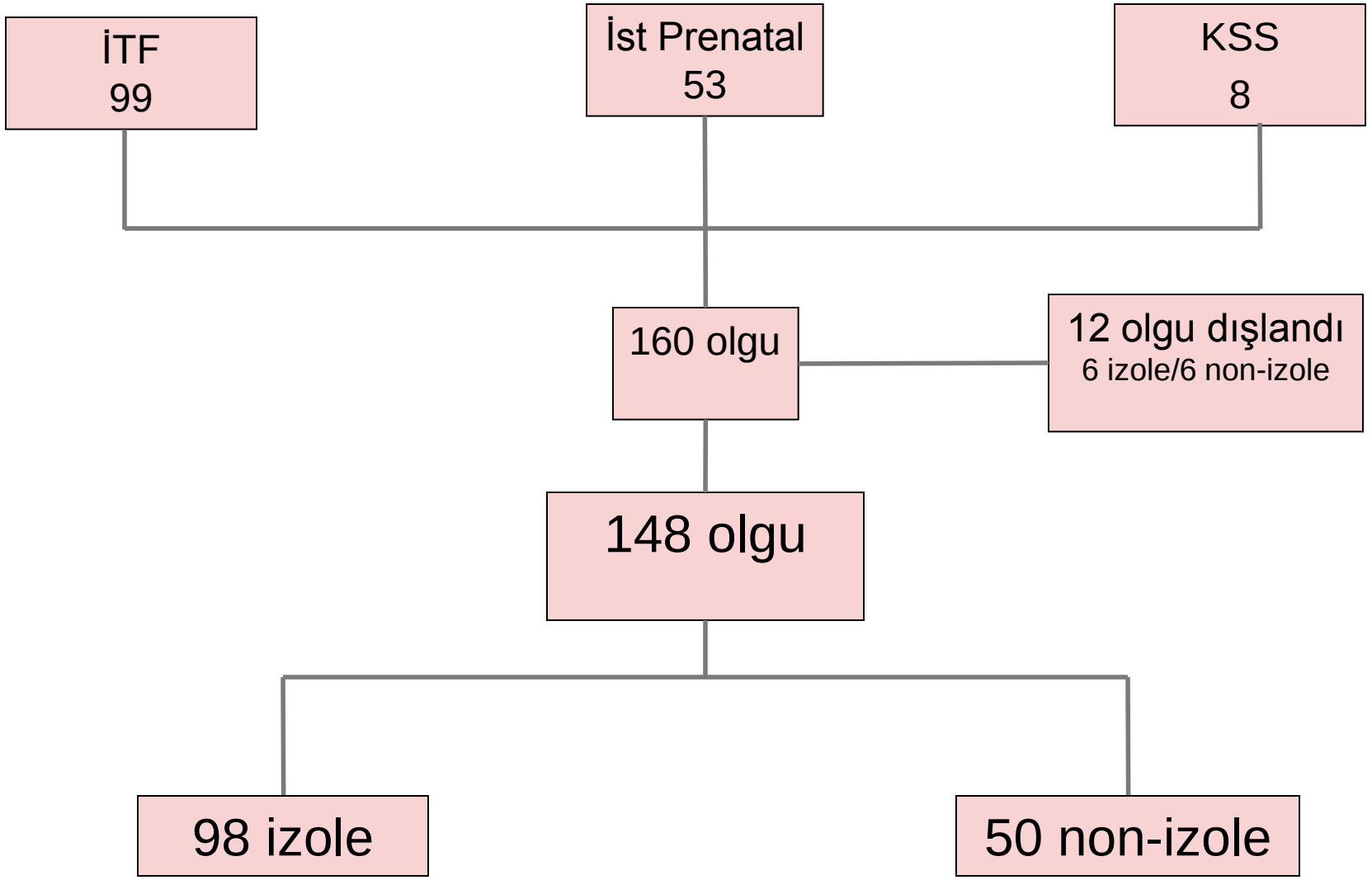
Gereç ve Yöntemler

- İzole / izole olmayan olgular
  - Kardiyak anomali
  - Ekstrakardiyak bulgu (FBK, anomalier)
  - Soft marker
    - Artmış ense plisi kalınlığı
    - Nazal kemik hipoplazisi
    - Ekojenik barsak
    - Ekojenik intrakardiyak odak
    - Tek umbilkal arter
    - Femor veya humerus uzunluğu <5. persantil
    - Koroid pleksus kisti
    - Hafif pelviektazi

Gereç ve Yöntemler

- Veriler hastane dosyalarından ve bilgisayar kayıtlarından elde edildi
- Takibe gelmeyen hastalar telefonla arandı
- İstatistiksel analizler için Ki-kare ve Mann-Whitney U test kullanıldı.

Bulgular


İTF
99
İst Prenatal
53
KSS
8
160 olgu
12 olgu dışlandı
6 izole/6 non-izole
148 olgu
98 izole
50 non-izole

Bulgular

- Tanı sırasında GH (medyan): 22 (14-34)
- 141 hastada invaziv karyotip analizi
- Kromozom anomalisi (T 21): %6.8 (10/148)
  - İzole: %6.1 (6/98)
  - İzole olmayan: %8 (4/50)

Bulgular

Table 1. Characteristics of cases with isolated and non-isolated ARSA

	Isolated (n = 98)	Non-isolated (n = 50)	Total (n = 148)
Median GA at diagnosis, weeks [min–max]	21 [16–30]	23 [14–34]	22 [14–34]
Median maternal age, years [min–max]	32 [22–50]	31 [19–46]	32 [19–50]
Maternal age ≥35	32 (32.7)	15 (30)	47 (31.8)
Trisomy 21	6 (6.1)	4 (8)	10 (6.8)
Outcome			
Normal clinical assessment	68 (69.4)	30 (60)	98 (66.2)
Termination of pregnancy	4 (4.1)	8 (16)	12 (8.1)
Postnatal ex	1 (1)[a]	1 (2)	2 (1.4)
Survival with Down syndrome	1 (1)	0 (0)	1 (0.7)
Survival after surgery	2 (2)[b]	1 (2)	3 (2)
Loss to follow-up	17 (17.3)	8 (16)	25 (16.9)
Not born yet	5 (5.1)	2 (4)	7 (4.7)

$p > 0.05$ for all parameters. Percentage values are shown in parentheses. [a] Postnatal ex due to preterm delivery at 24 weeks' gestation. [b] Surgery due to postnatally diagnosed anal stenosis and craniosynostosis.

Bulgular

- Ek kardiyak bulgu: %5.4 (8/148)
- Ekstrakardiyak bulgu: %10.8 (16/148)
- Soft marker: %24.3 (36/148)
- Sadece soft marker: %19.6 (29/148)

Bulgular

Table 3. Cardiac anomalies, extracardiac findings, and soft markers associated with ARSA (some of the fetuses had more than one abnormal finding)

Cardiac anomalies	n (%)	Extracardiac findings	n (%)	Soft markers	n (%)
PLSVC	3 (2)	fetal growth restriction	7 (4.7)	intracardiac echogenic focus	14 (9.5)
Rhabdomyoma	1 (0.7)	cleft lip palate	2 (1.3)	mild pyelectasis	12 (8.1)
Aortic stenosis	1 (0.7)	arachnoid cyst	2 (1.3)	single umbilical artery	5 (3.4)
Dextrocardia	1 (0.7)	cystic hygroma	2 (1.3)	nasal bone hypoplasia	5 (3.4)
Ventricular septal defect	1 (0.7)	corpus callosum agenesis	1 (0.7)	short femur and humerus	3 (2)
Dilated ductus arteriosus	1 (0.7)	thymic aplasia	1 (0.7)	choroid plexus cyst	2 (1.3)
		PRUV	1 (0.7)	fetal echogenic bowel	1 (0.7)
		hydrops	1 (0.7)	thick nuchal fold	1 (0.7)
		polydactyl	1 (0.7)		
		cloacal dysgenesis	1 (0.7)		
		hydrocephaly	1 (0.7)		
		hydrometrocolpos	1 (0.7)		
		holoprosencephaly	1 (0.7)		
		hypertelorism	1 (0.7)		
		omphalocele	1 (0.7)		
		umbilical cord angioma	1 (0.7)		

PLSVC = Persistent left superior vena cava; PRUV = persistent right umbilical vein.

Bulgular

Table 4. Characteristics of cases with ARSA and trisomy 21

Case	Maternal age, years	GAD, weeks	Screening test result	Cardiac finding	Extracardiac finding	Soft marker	Outcome
1	37	22	NP	none	none	none	alive
2	39	19	positive	none	none	none	TOP
3	29	19	positive	none	none	none	TOP
4	38	20	NP	none	none	none	TOP
5	42	17	positive	none	none	none	unknown
6	35	16	positive	none	none	none	TOP
7	42	21	NP	none	hydrops fetalis	NBH, NF, CPC	TOP
8	26	24	NP	none	FGR	SUA, FEB	postnatal ex
9	30	21	negative	none	FGR	pyelectasis, EIF, NBH	TOP
10	41	16	positive	none	none	EIF	TOP

GAD = Gestational age at diagnosis; NP = not performed; TOP = termination of pregnancy; FGR = fetal growth restriction; NBH = nasal bone hypoplasia; NF = thick nuchal fold; CPC = choroid plexus cyst; SUA = single umbilical artery; FEB = fetal echogenic bowel; EIF = intracranial echogenic focus.

6/10 izole
Soft marker: 4
FBK: 2
Hidrops:1
Kardiyak anomali: 0

Sonuç

- ARSA olgularında kardiyak ve ekstrakardiyak anomali olasılığı nedeniyle detaylı anomali taraması şart.
- İzole ARSA olgularının %6.1'inde T 21
- İzole ve izole olmayan ARSA olgularında T21 insidansı anlamlı olarak farklı değil (%6.1 vs. %8)
- ARSA + T 21 olgularının %60'ında ARSA tek bulgu

Sonuç

	Izole ARSA	Toplam	%
Chaoui et al. (2005)[1]	1	5	20
Zalel et al. (2008)[5]	0	3	0
Borenstein et al (2010)[4]	1	12	8.3
Rembouskos et al.(2010)[8]	1	6	16.6
Paladini et al. (2012)[9]	8	27	29.6
Willruth et al. (2012)[3]	0	1	0
Yazicioglu et al. (2013)[10]	0	7	0
Bu çalışma	6	10	60

ARSA'ya eşlik eden bulgu olmasa da karyotip analizi yapmak mantıklı

Fetal Diagnosis and Therapy

Original Paper

Fetal Diagn Ther
DOI: 10.1159/000354650

Received: March 25, 2013
Accepted after revision: July 23, 2013
Published online: September 14, 2013

Detection Rate of Trisomy 21 in Fetuses with Isolated and Non-Isolated Aberrant Right Subclavian Artery

Aytul Corbacioglu Esmer[a] Ahmet Gul[b] Aslı Nehir[a] Atil Yuksel[a] Ozlem Dural[c]
Ibrahim Kalelioglu[a] Recep Has[a] Tanju Demiroren[d]

TEŞEKKÜRLER…