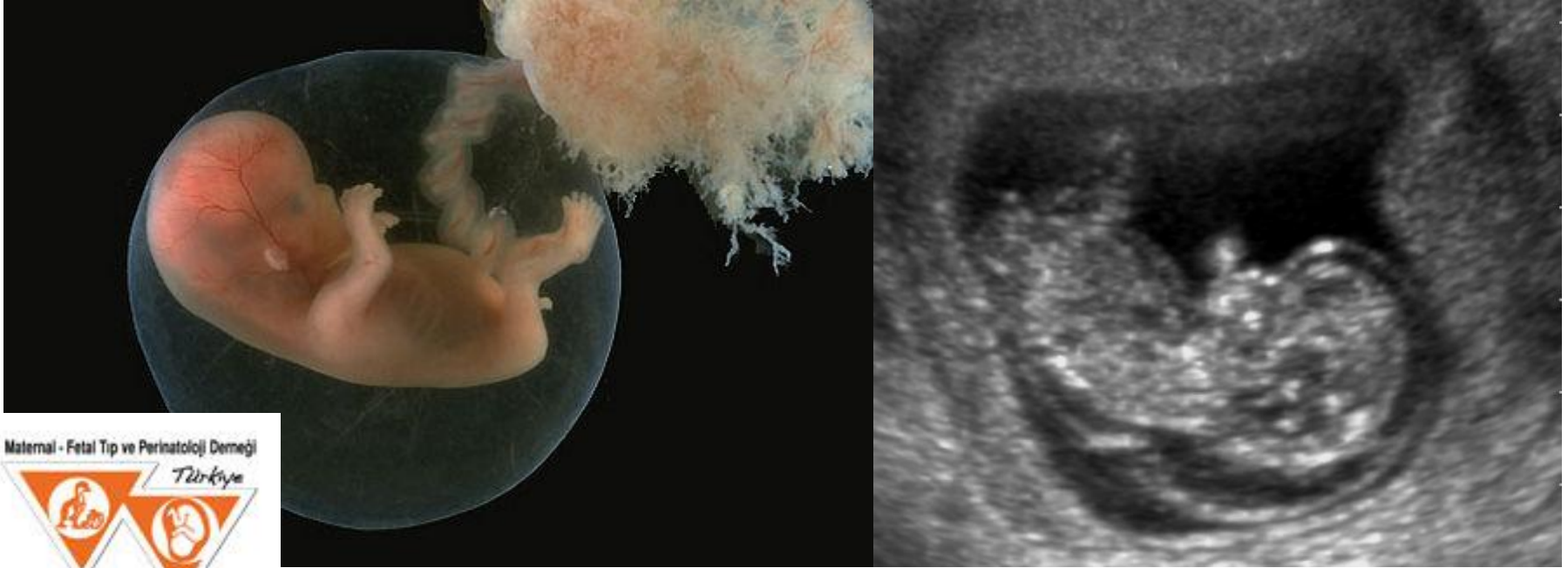


# BİRİNCİ TRİMESTER ULTRASONOGRAFİK ANÖPLOİDİ BELİRTEÇLERİ



Maternal - Fetal Tıp ve Perinatoloji Derneği



PERİNATOLOJİ  
UZMANLARI  
DERNEĞİ

Prof.Dr. **FİLİZ F. BİLGİN YANIK**

# KROMOZOM ANOMALİLERİ

## Sayısal anomaliler

Poliploidiler

Triploidi=3n

Tetraploidi=4n

Anöploidiler

Trizomi 21, 18, 13

Monozomi X

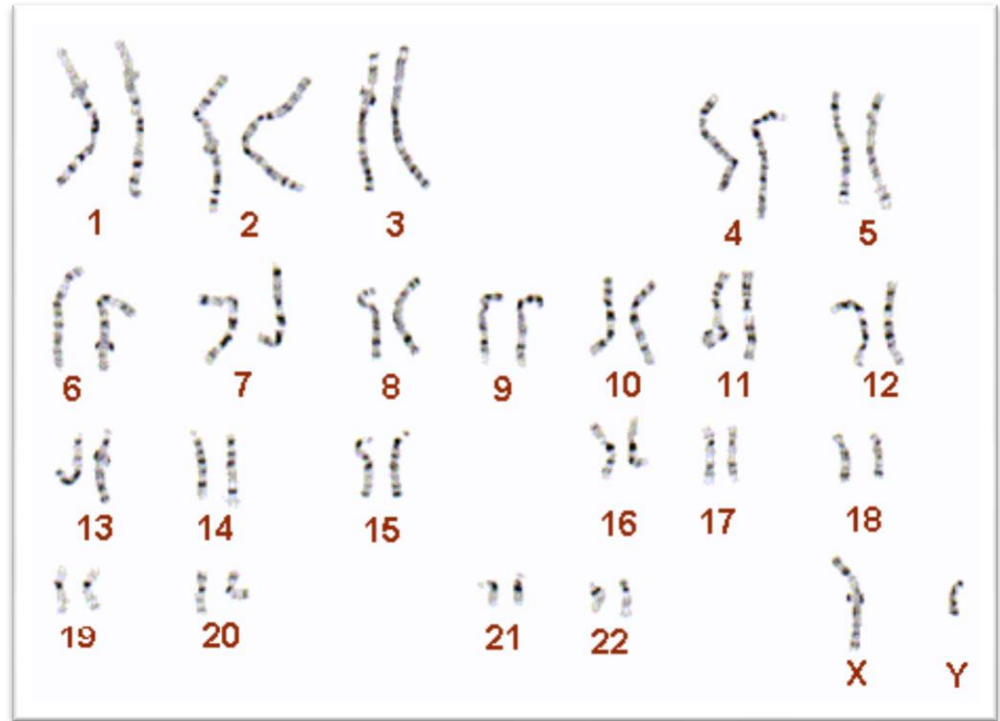
Markır kromozom

## Yapısal anomaliler

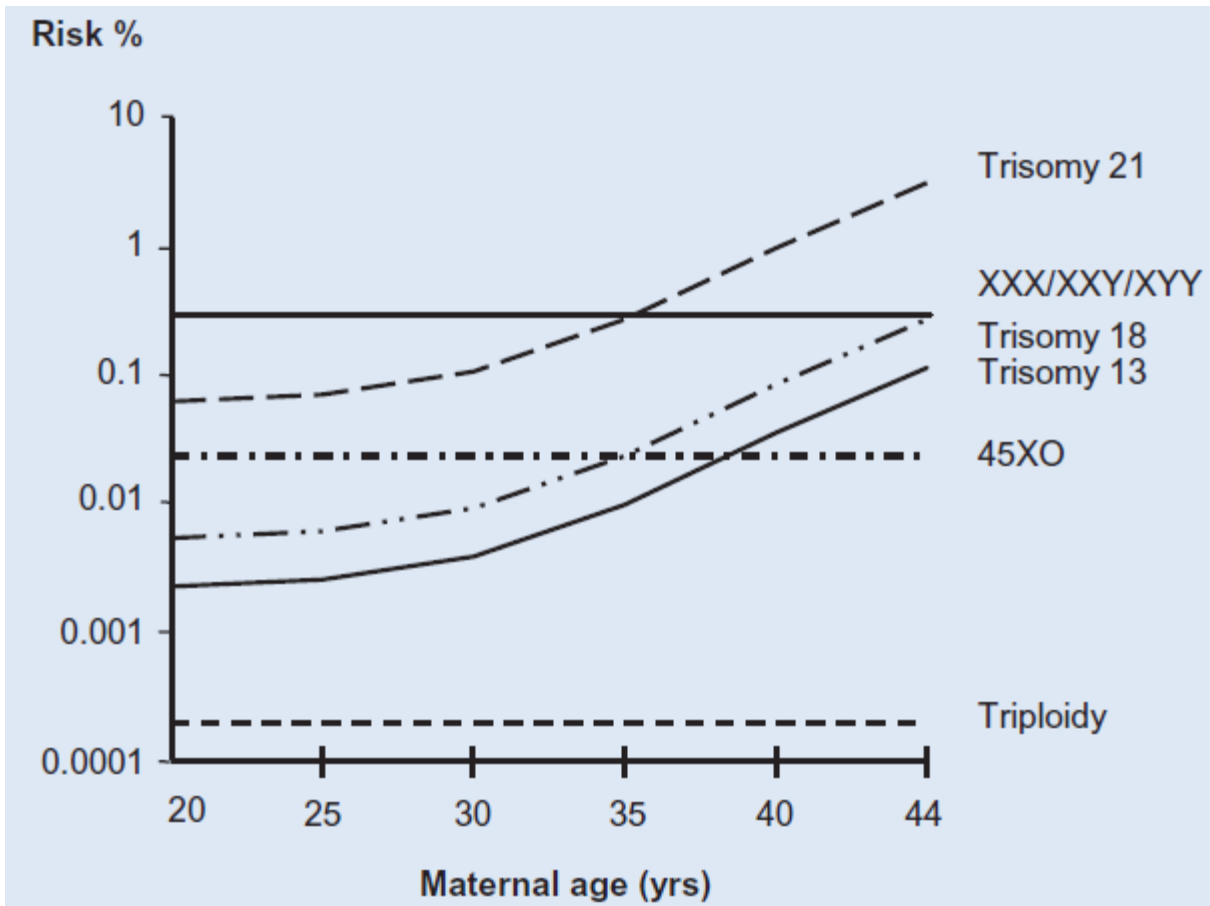
İnversiyonlar

Delesyonlar

Translokasyonlar



# ANNE YAŞI VE KROMOZOM ANOMALİLERİ İLİŞKİSİ



# ANNE YAŐI VE KROMOZOM ANOMALİLERİ İLİŐKİSİ

Yaő	Down Sendromu	Kromozom Anomalileri
20	1/1429-1/2000	1/526
25	1/1111-1/1429	1/476
30	1/833-1/1111	1/385
35	<b>1/256-1/400</b>	<b>1/179</b>
40	<b>1/73-1/118</b>	<b>1/63</b>
45	<b>1/19-1/35</b>	<b>1/18</b>

Ernest Hook. Age and chromosome abnormalities.  
Obstetrics and Gynecology, September 1981.

# TARAMA TESTLERİ DS SAPTAMA ORANLARI

(%5 Yalancı Pozitiflik)

- Anne yaşı %30
- 1.TMBT %60
- **1.TM NT %75**
- 1.TM Kombine Test %85
- **1.TM Kombine Test+USG >%90**
- 2.TMBT (üçlü-dörtlü) %60-80
- Entegre/Serum Entegre Test %90
- sfDNA (<%1 yalancı pozitiflik) %99

# I. TM ARTMIŞ ENSE SAYDAMLILIĞI (Nuchal Translucency-NT)



- 11-13+6 hf / CRL 45-84 mm
- Zum (fetal baş ve toraks)
- Sagital kesit- baş nötr
- Kaliperler çizgilerin üstünde ve iç tarafta olmalı - maksimum ölçüm alınmalı
- Artmış NT: septalı/septasız, ense arkasında/yaygın olabilir



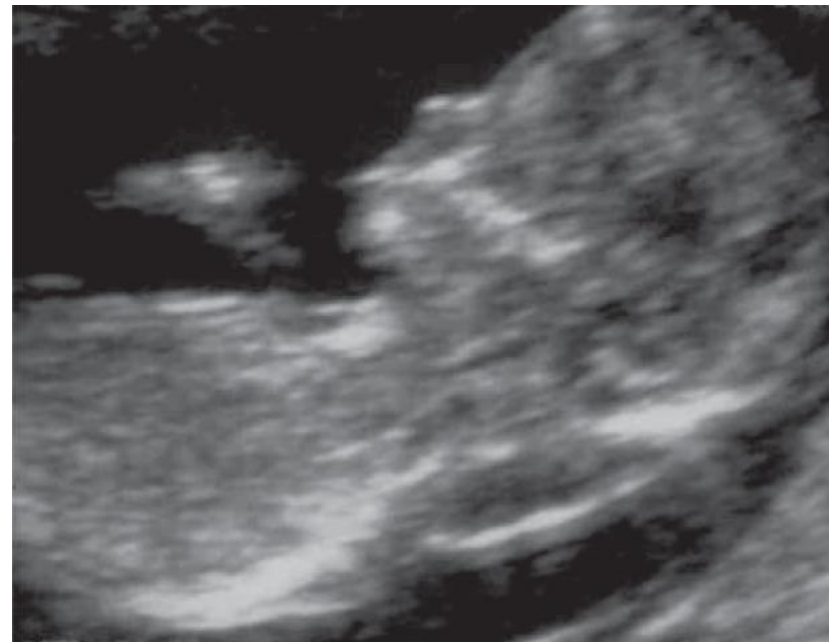
# I. TRİMESTER ENSE SAYDAMLIGI

Artmış NT  
 $\geq 1,5$  MoM / > 95. Persantil

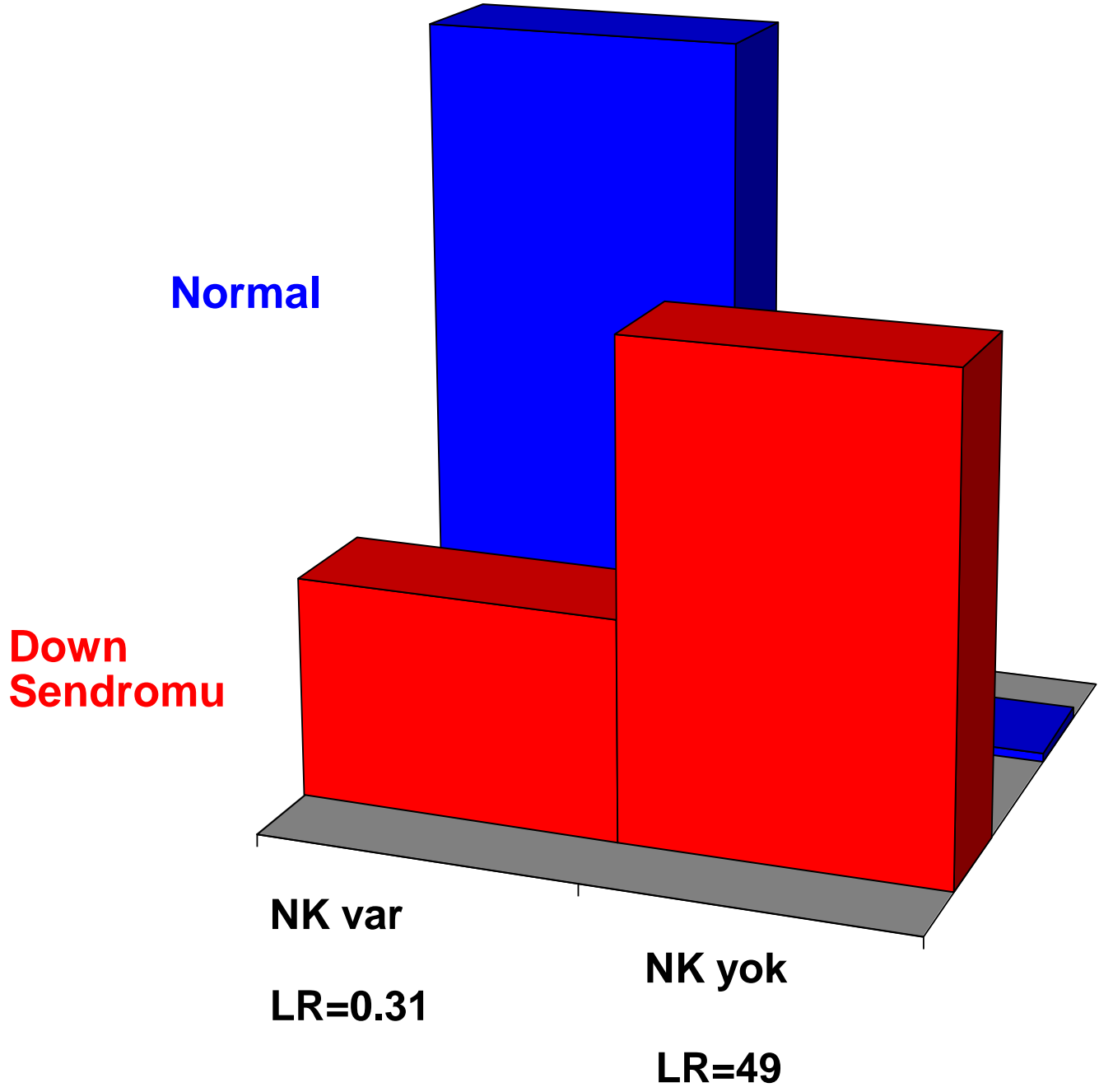
- Kromozom anomalileri (T21,T18,T13,Turner S,vb)
- Genetik Sendromlar (Noonan S,vb)
- Fetal yapısal anomaliler (Kardiyak anomaliler, konjenital diafragma hernisi,iskelet displazileri,vb)
- Fetal lenfatik sistem gelişiminde problemler
- Fetal nöro-musküler problemler
- Fetal anemi
- Konjenital enfeksiyonlar
- İİTS

# I.TM USG - Nazal Kemik Yokluđu

- NK yokluđu → DS:%60-70  
T 18:%50  
T 13:%30  
Normal fetuslarda: %2







Normal

Down  
Sendromu

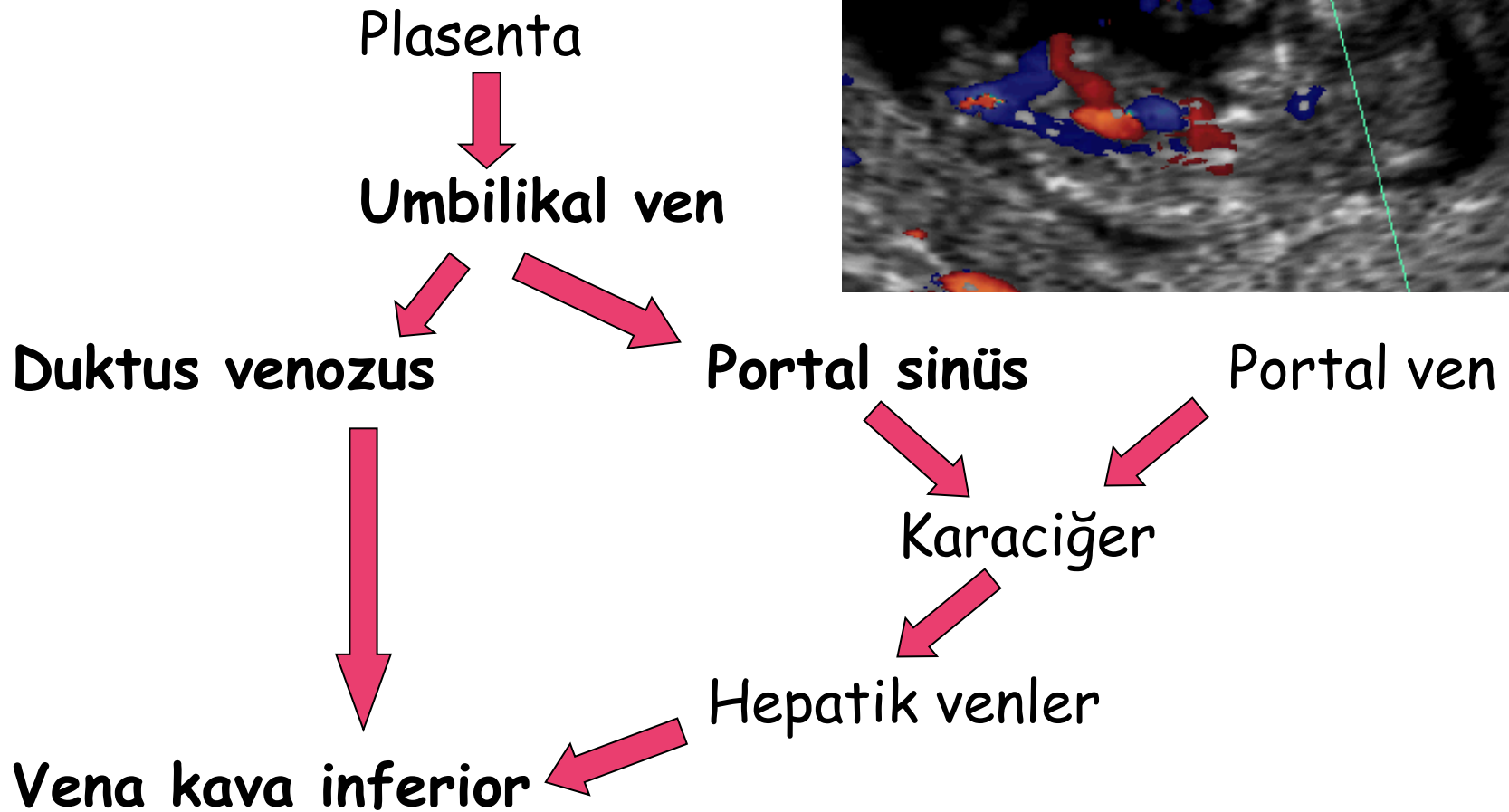
NK var

LR=0.31

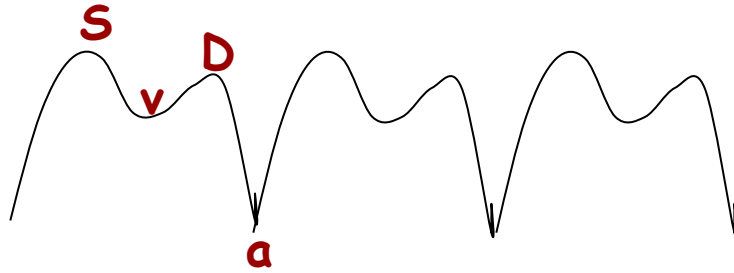
NK yok

LR=49

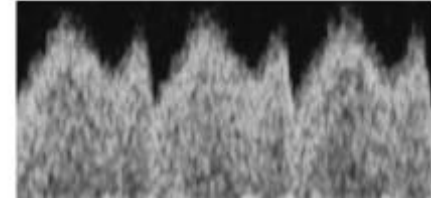
# I. TM USG - DVD



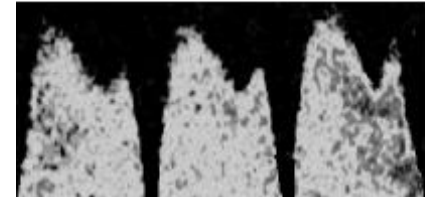
# I. TM USG - DVD



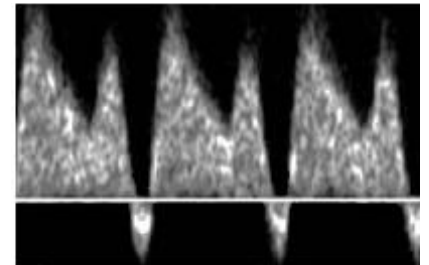
- S: ventriküler sistol
- D: Ventriküler diastol
- a: Atrial sistol



normal



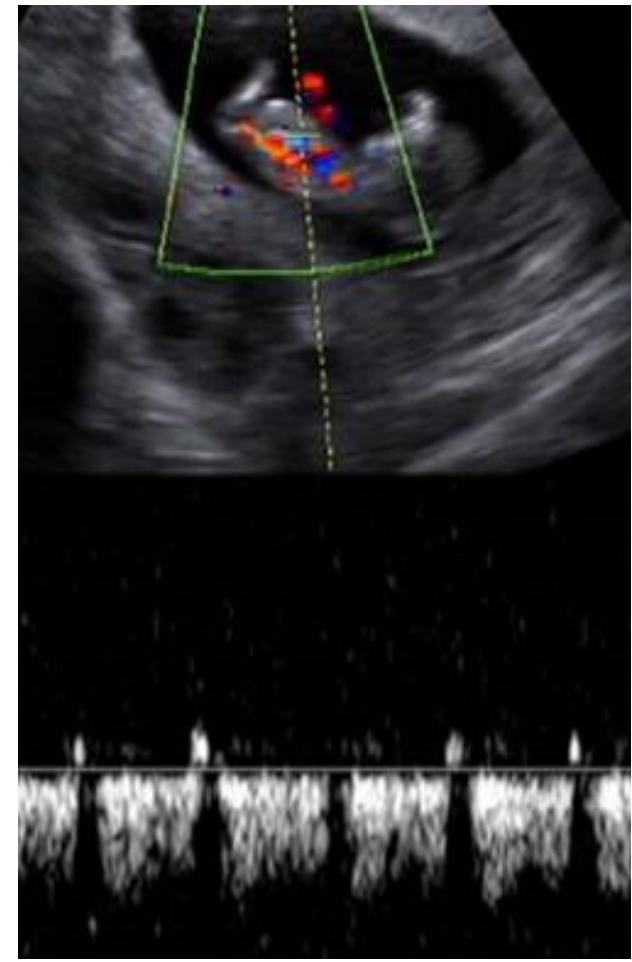
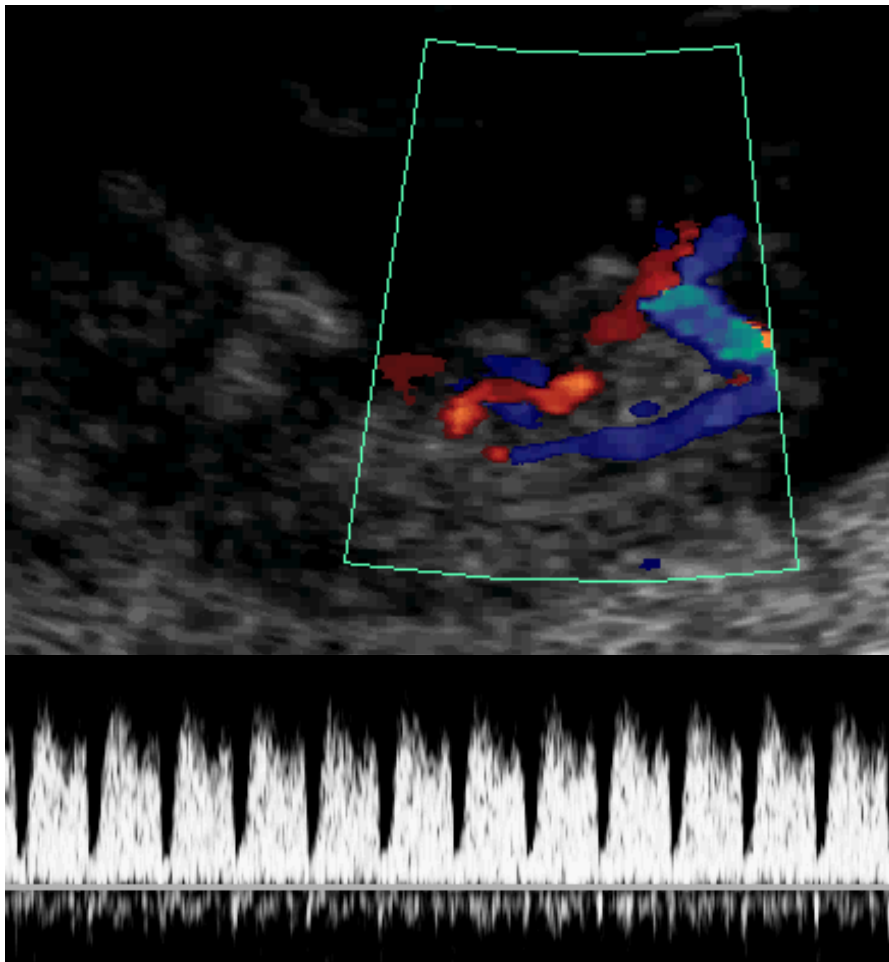
a dalgası kaybı



ters a dalgası

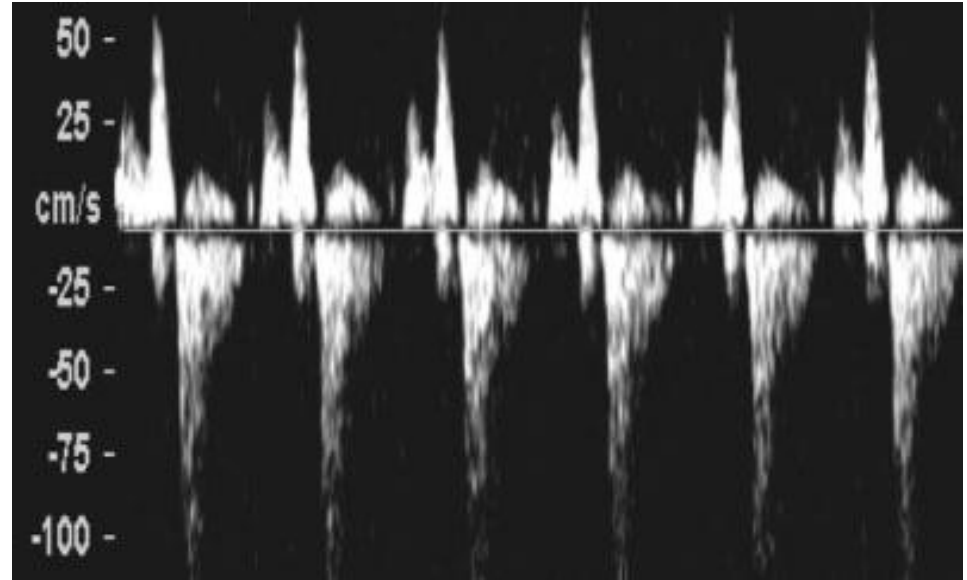
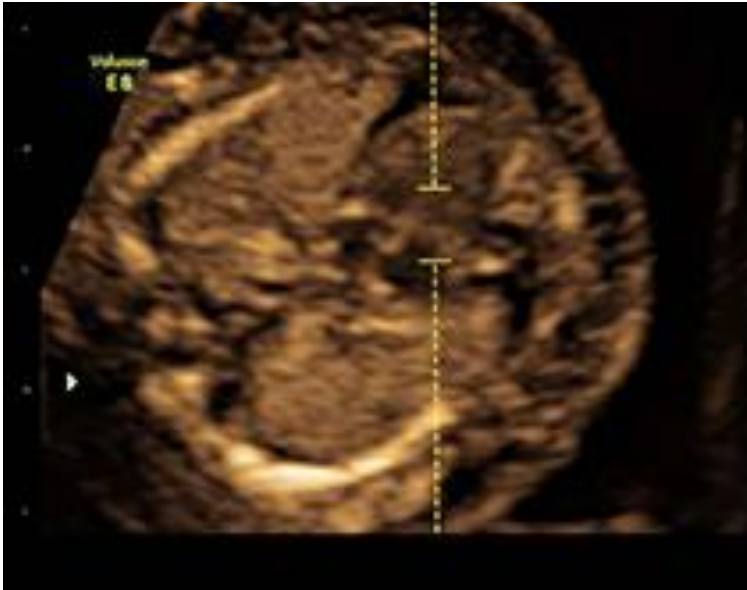
# I.TM USG - Duktus Venozus Doppleri

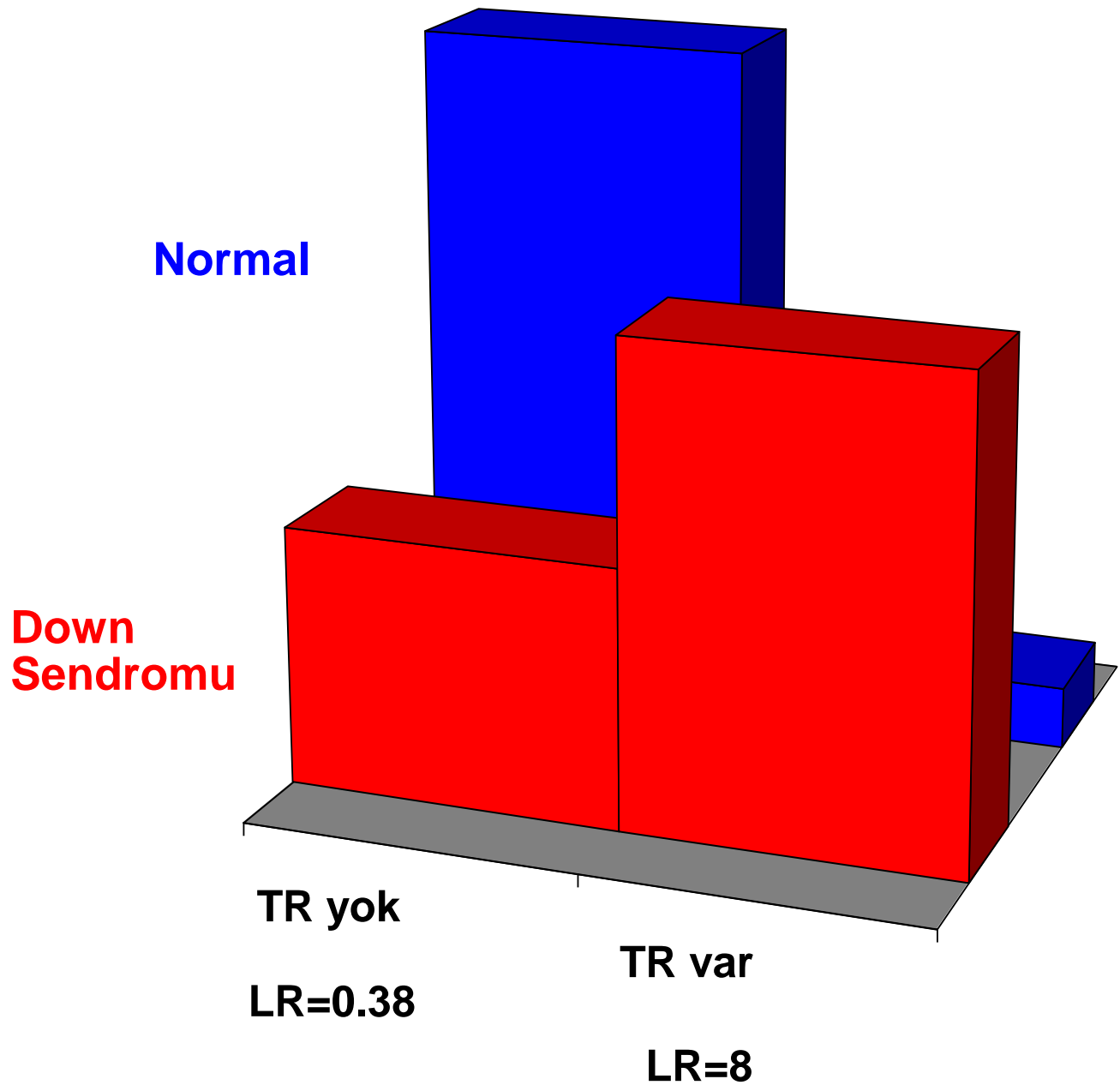
- Anormal DVD → DS:%80  
Normal fetuslarda: %5



# I.TM USG - Triküspid Regürjidasyon

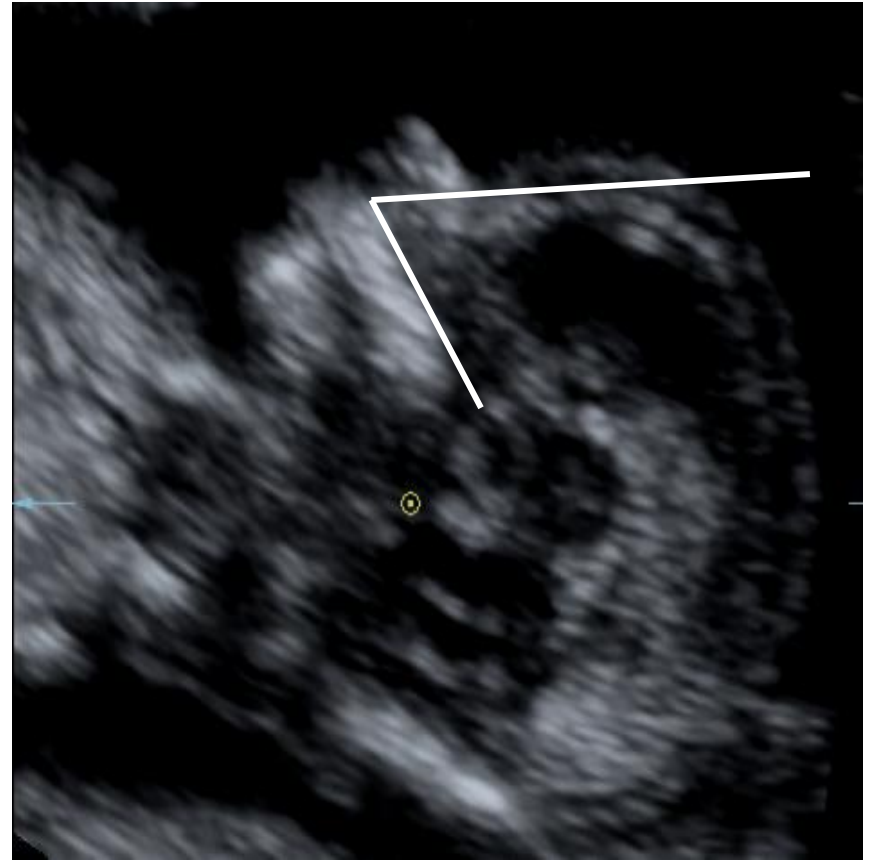
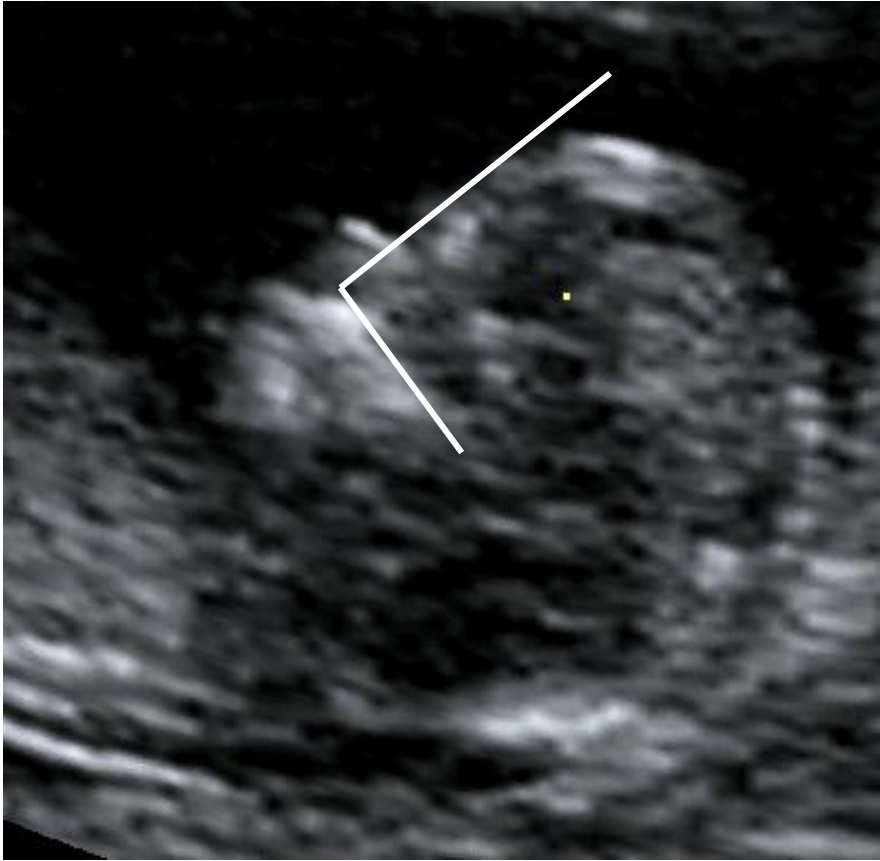
- Triküspid Regürjidasyon → DS:%55  
Normal fetuslarda:%1



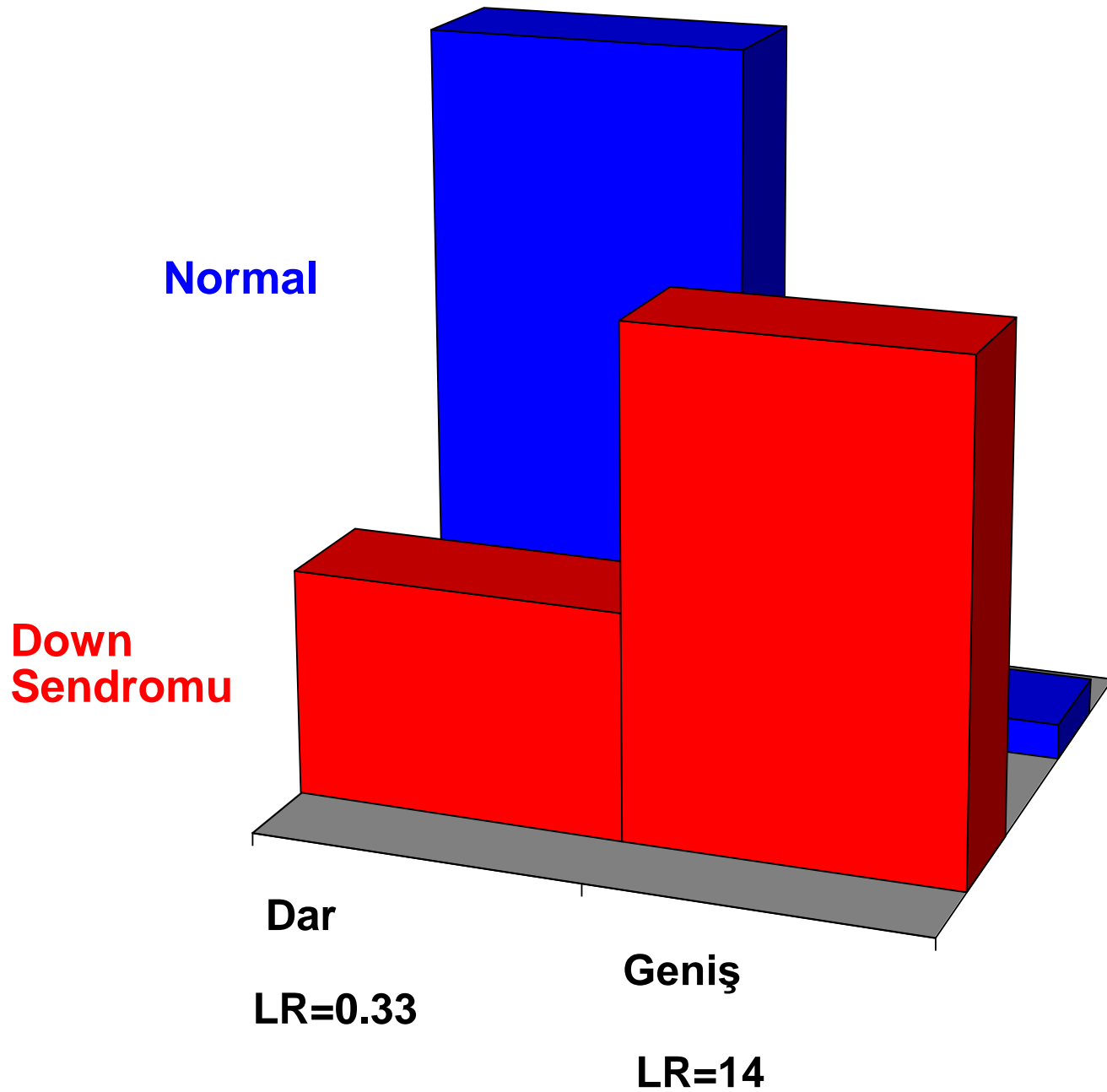




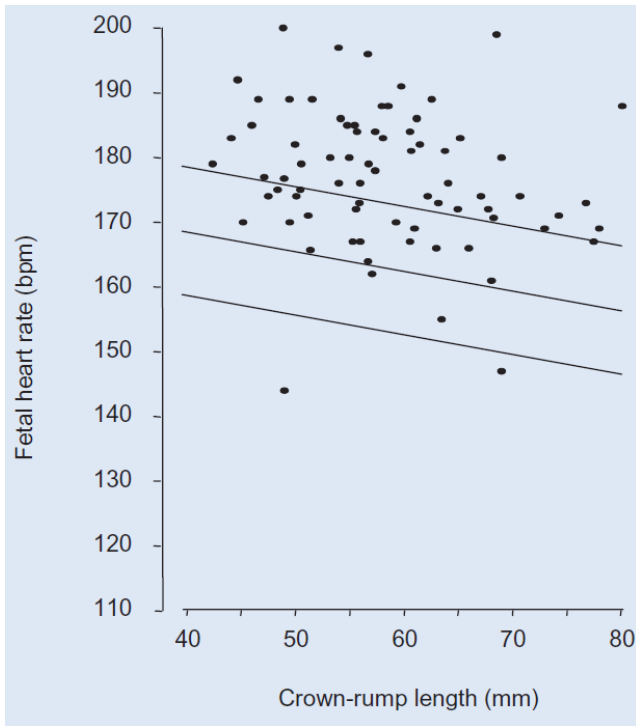
# I.TM USG - Frontomaksiller Fasiyal Açı



- Normal fetuslarda  $82,9^{\circ} \pm 4,1^{\circ}$
- Down Sendromunda  $92,3^{\circ} \pm 5,2^{\circ}$



# I.TM USG - Fetal Kalp Atım Hızı



10 → 14 hf

170 → 155/dk

➤ T13, Turner Sendromu  
Taşikardi

➤ T18, Triploidi  
Bradikardi

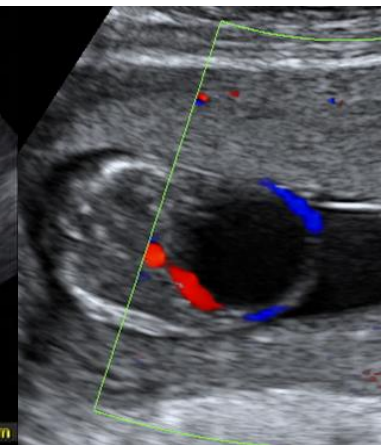
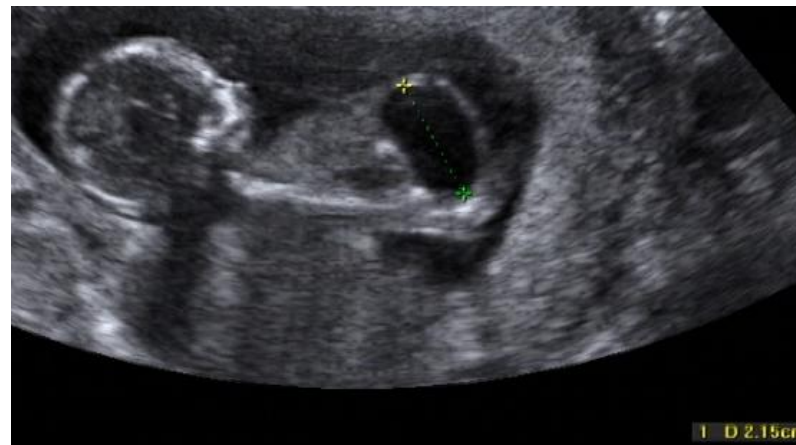
# I.TM USG - Trizomi 18

- Artmış NT
- NK yokluğu
- Erken büyüme kısıtlılığı\*
- Bradikardi
- Omfalosel %60 (N:1/1000)
- TUA %80 (N:%3)
- Megacistis (özellikle 7-15mm) %20 (N:1/1500)



# I. TM USG - Trizomi 13

- Artmış NT
- NK yokluğu
- Erken büyüme kısıtlılığı
- Taşikardi
- Megacistis
- Holoprozensefali
- Omfalosel



# I. TM USG - Turner Sendromu

- Artmış NT
- Erken büyüme kısıtlılığı
- Taşikardi



# I. TM USG - Triploidi

- Asimetrik erken büyüme kısıtlılığı\*
- Bradikardi
- Holoprozensefali
- Omfalosel
- Posterior fossa kisti
- Plasentada molar değişiklikler



# I. TRİMESTER ANÖPLOİDİ TARAMASI

Anne yaşı + NT+ 1.TM BK

+ Nazal kemik + DV Doppleri + TR

DS saptama oranı >%90 (%5 yalancı pozitiflik)

Kombine riski 1/50-1/1000 arasında olan olgularda  
CVS kararında önemli \*\*\*

