Birinci trimester taramasında güncel durum

Prof. Dr. Hayri Ermiş anısına…

Birinci trimester tarama

Anne Yaşı +

- Ultrason
  - CRL
  - NT
  - NK
  - Duktus venosus
  - Triküspit regurjitasyonu
  - Diğer...
- Biyokimya
  - Serbest beta hCG
  - PAPP-A
  - PLGF
  - AFP

Fetal nuchal translucency: ultrasound screening for chromosomal defects in first trimester of pregnancy

K H Nicolaides, G Azar, D Byrne, C Mansur, K Marks

BMJ. 1992 April 4; 304(6831): 867–869.

Abstract

***Objective*—To examine the significance of fetal nuchal translucency at 10-14 weeks' gestation in the prediction of abnormal fetal karyotype.**

***Design*—Prospective screening study.**

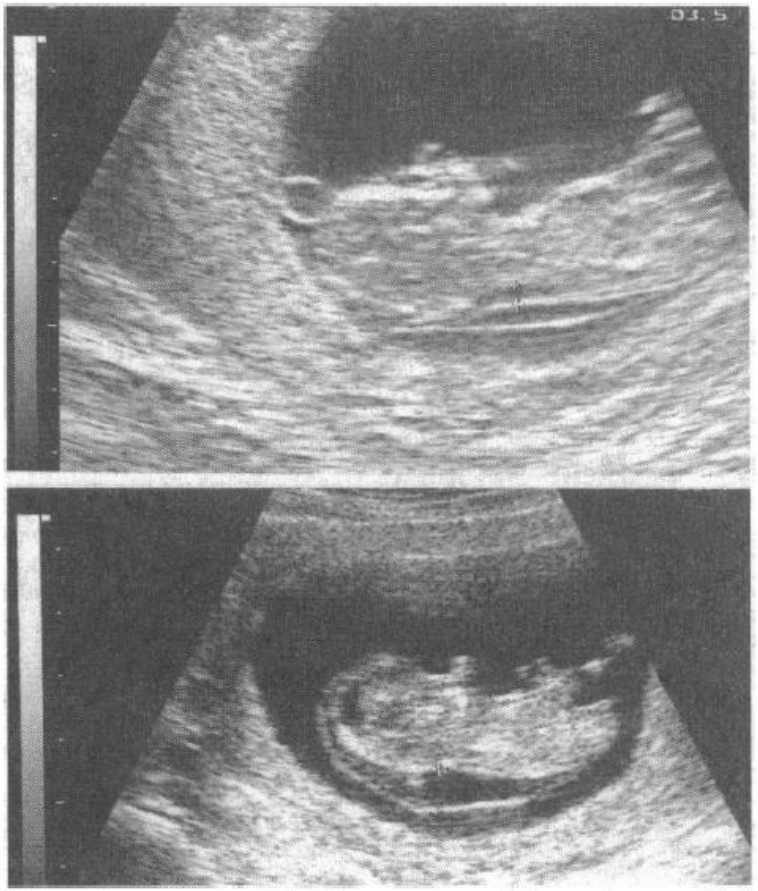
***Setting*—The Harris Birthright Research Centre for Fetal Medicine, King's College Hospital, London.**

***Subjects*—827 fetuses undergoing first trimester karyotyping by aminocentesis or chorionic villus sampling.**

***Main outcome measure*—Incidence of chromosomal defects.**

***Results*—The incidence of chromosomal defects was 3% (28 of 827 cases). In the 51 (6%) fetuses with nuchal translucency 3–8 mm thick the incidence of chromosomal defects was 35% (18 cases). In contrast, only 10 of the remaining 776 (1%) fetuses were chromosomally abnormal.**

***Conclusion*—Fetal nuchal translucency ≥3 mm is a useful first trimester marker for fetal chromosomal abnormalities.**

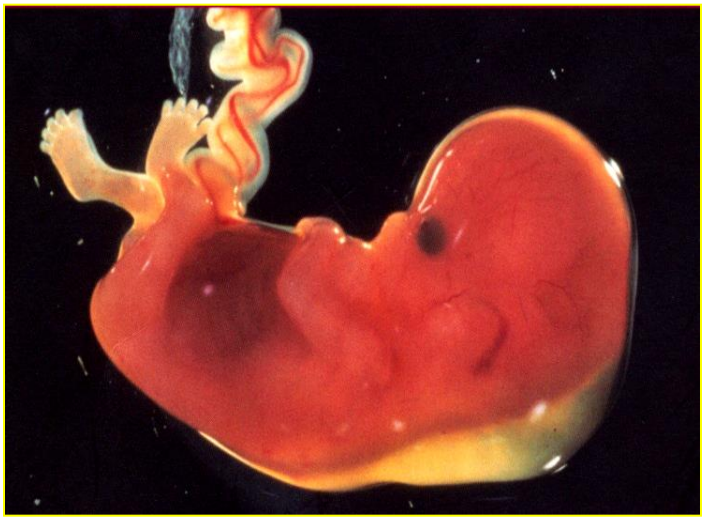


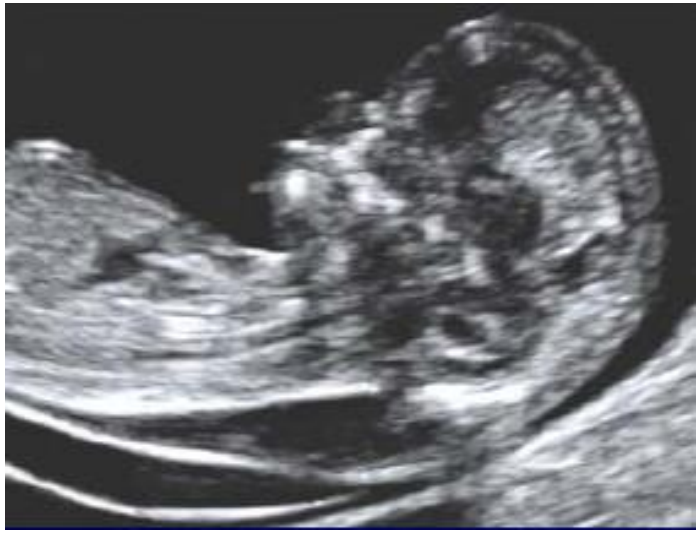
Ultrasonic appearances of subcutaneous nuchal translucency. Both skin and amnion appear as thin membranes (top). In some cases translucency extends over wide area of fetus but is most prominent behind neck (bottom)

Introduction

In the second and third trimesters of pregnancy there is a high association between fetal nuchal cystic hygromas or nuchal oedema and chromosomal

Artmış NT

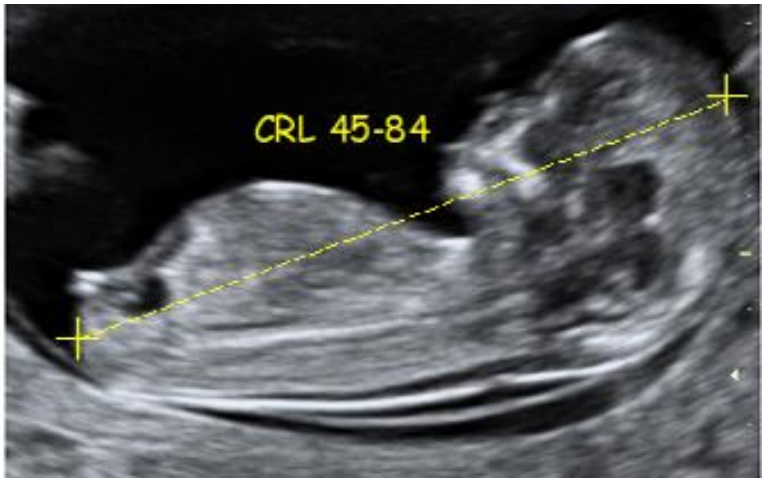


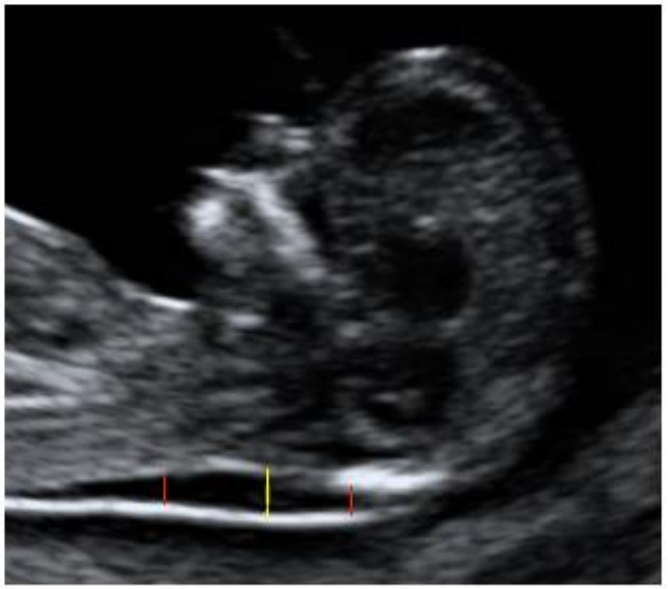


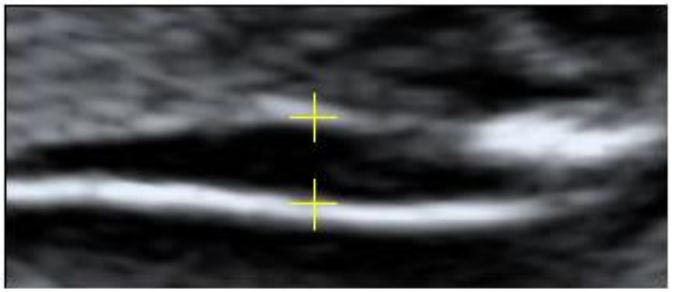
- Kardiyak anomaliler / yetmezlik
- Intratorasik kompresyon
- Anormal lenfatik sistem
- Nöro-musküler anomaliler
- Kollagen sentezinde değişiklikler

“Nuchal Translucency”

- Mid-sagittal kesit
- Büyük görütü
- Nötral pozisyon
- Amniyon zarı ayrık
- Kaliper: 0.1mm
- Kalliperler “on-to-on







11+0-13+6 hf

‘NT’ çalışma sonuçları

Authors	n	*Gestation (weeks)*	*NT cut-off*	*FPR (%)*	*Trisomy 21 DR (%)*
Pandya *et al.* (1995)[7]	1763	10–14	≥2.5 mm	3.6	3/4 (75)
Szabo *et al.* (1995)[8]	3380	9–12	≥3.0 mm	1.6	28/31 (90)
Taipale *et al.* (1997)[9]	6939	10–14	≥3.0 mm	0.8	4/6 (67)
Hafner *et al.* (1998)[10]	4371	10–14	≥2.5 mm	1.7	4/7 (57)
Pajkrt *et al.* (1998)[11]	1547	10–14	≥3.0 mm	2.2	6/9 (67)
Snijders *et al.* (1998)[12]	96 127	10–14	≥95th centile	4.4	234/327 (72)
Economides *et al.* (1998)[13]	2281	11–14	≥99th centile	0.4	6/8 (75)
Schwarzler *et al.* (1999)[14]	4523	10–14	>2.5 mm	2.7	8/12 (67)
Theodoropoulos *et al.* (1998)[15]	3550	10–14	≥95th centile	2.3	10/11 (91)
Zoppi *et al.* (2001)[16]	12 311	10–14	≥95th centile	5.0	52/64 (81)
Gasiorek-Wiens *et al.* (2001)[17]	23 805	10–14	≥95th centile	8.0	174/210 (83)
Brizot *et al.* (2001)[18]	2996	10–14	≥95th centile	5.3	7/10 (70)
Audibert *et al.* (2001)[19]	4130	10–14	≥95th centile	4.3	9/12 (75)
Wayda *et al.* (2001)[20]	6750	10–12	≥2.5 mm	4.3	17/17 (100)
Totals	174 473			4.7	562/728 (77)

DR, detection rate; FPR, false-positive rate.

Yakalama oranı: %77

NT ve Diğer kromozom anomalileri

Karyotip	n	NT>95.per	➢1/300
Trizomi 18	119	89	%81
Trizomi 13	46	33	%80
Turner	54	47	%89
Triploidi	32	19	%62
Diğer	74	41	%79
Toplam	325	229	%78

Snijders RJM. Lancet 1998

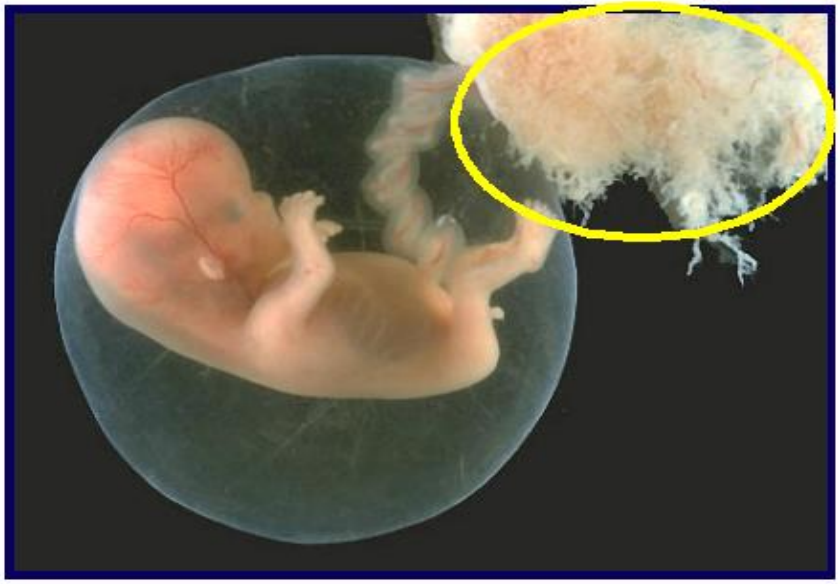
Screening for fetal chromosomal abnormalities with nuchal translucency measurement in the first trimester.

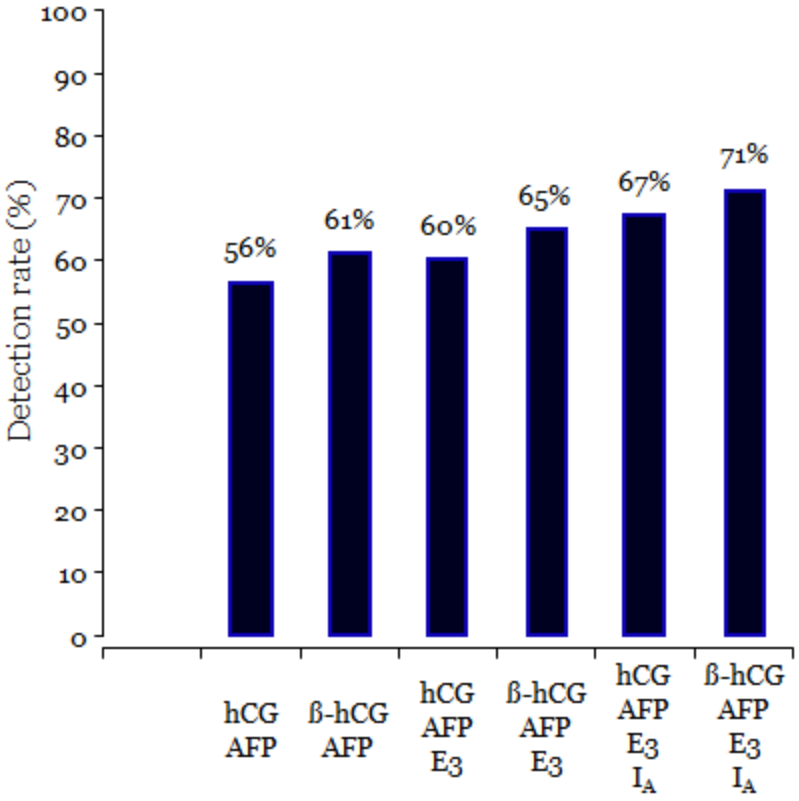
Has R, Kalelioglu I, Ermis H, Ibrahimoglu L, Yuksel A, Yildirim A, Basaran S.
Fetal Diagn Ther. 2006;21(4):355-9

N: 4,598 fetus (4,365 gebelik)
32 Kromozom anomalisi
Down:19
Diğer: 13

Kromozom Anomalisi	Kromozom Anomalisi Yakalama Oranı
	Maternal Yaş + NT (1/300 - %4,7 YPO ile)
Trizomi 21	**14/19 (%73.6)**
Trizomi 18	4/5 **(%80)**
Tümü	22/32 **(%68,8)**

Biyokimyasal tarama

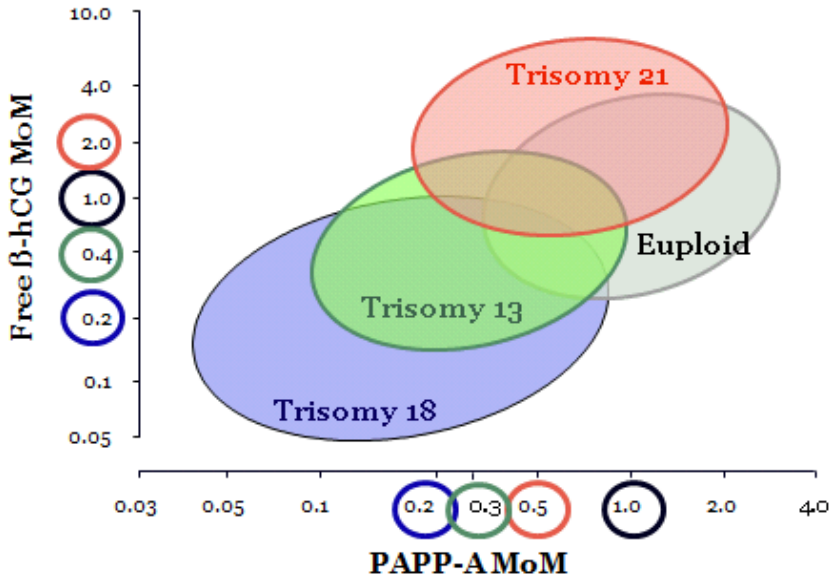




H Cuckle, P Penn, D Wright, Semin Perinatol 2005;29:252-7

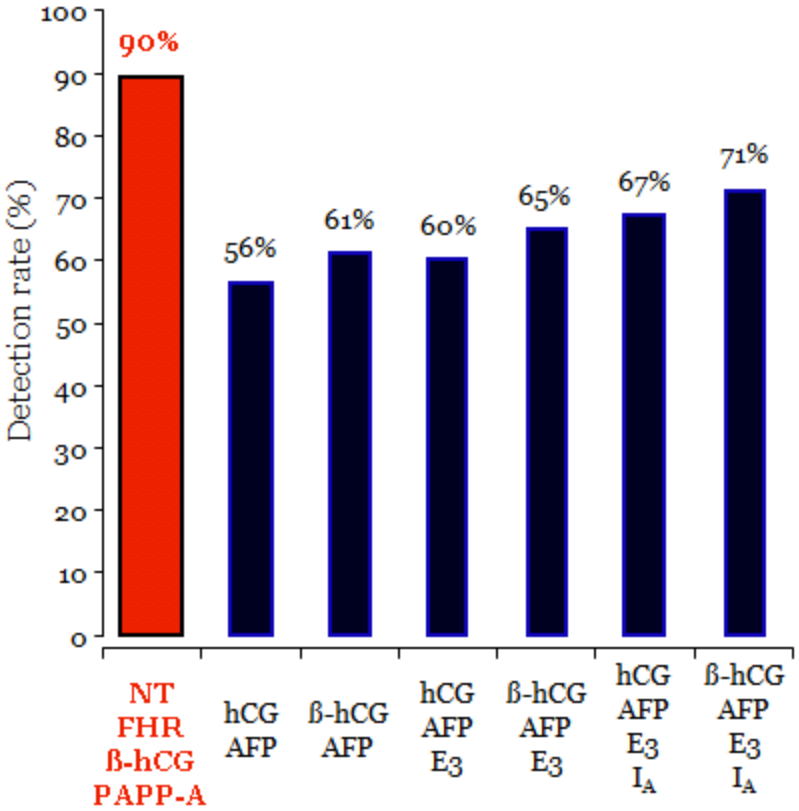
free ß-hCG PAPP-A

	free ß-hCG	PAPP-A
Trizomi 21	2.0	0.5
Trizomi 18	0.2	0.2
Trizomi 13	0.3	0.4
Turner	1.2	0.5
Triploidi		
Digynik	0.2	0.1
Diandrik	9.0	0.7



Birinci trimester taraması

- Kombine Test
  - Maternal yaş
  - Fetal NT
  - Serum free ß-hCG+
  - PAPP-A
- Trizomi 21
  - DR: %90
  - FPR: %3

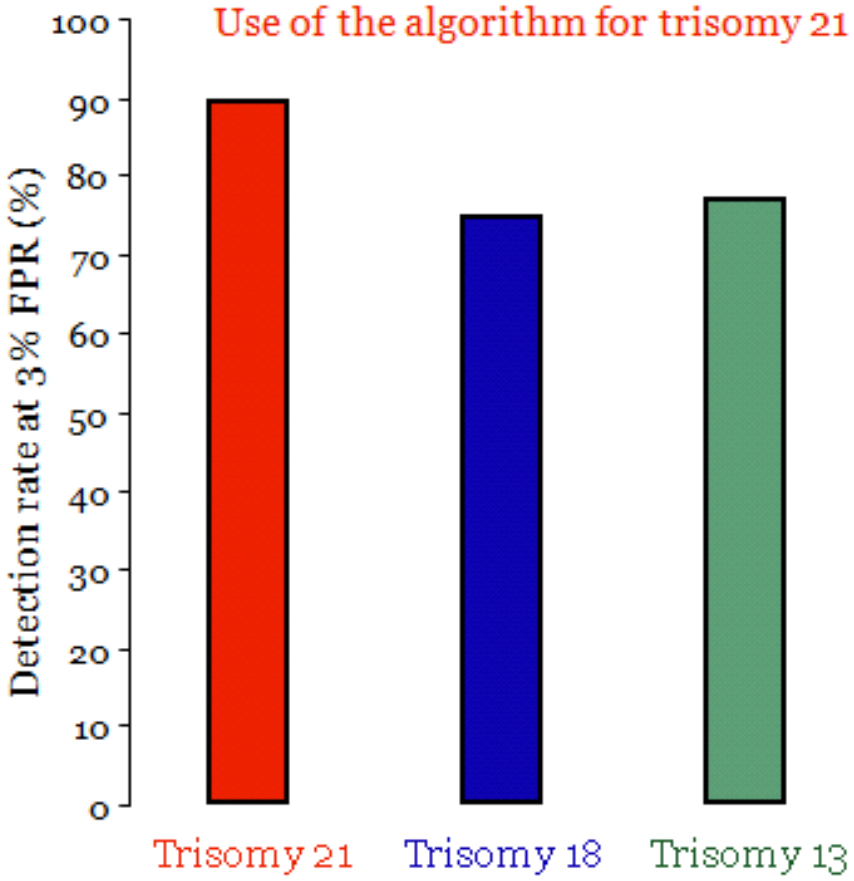


H Cuckle, P Penn, D Wright, Semin Perinatol 2005;29:252-7

Birinci trimester taraması

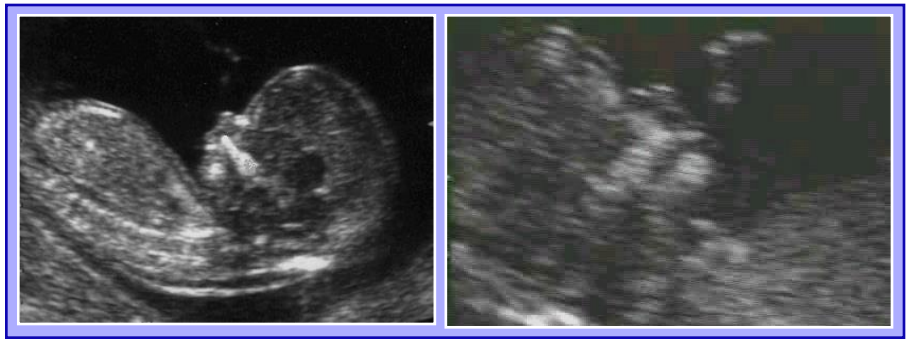
- Trizomi 18
- Trizomi 13

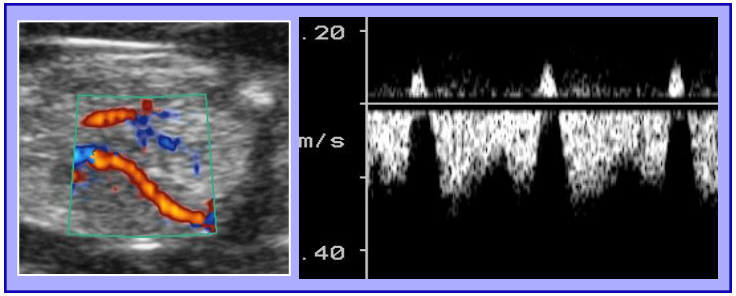
  - **DR: %75 FPR: %3**

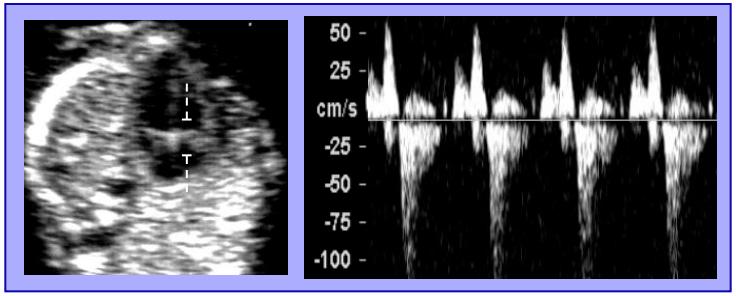


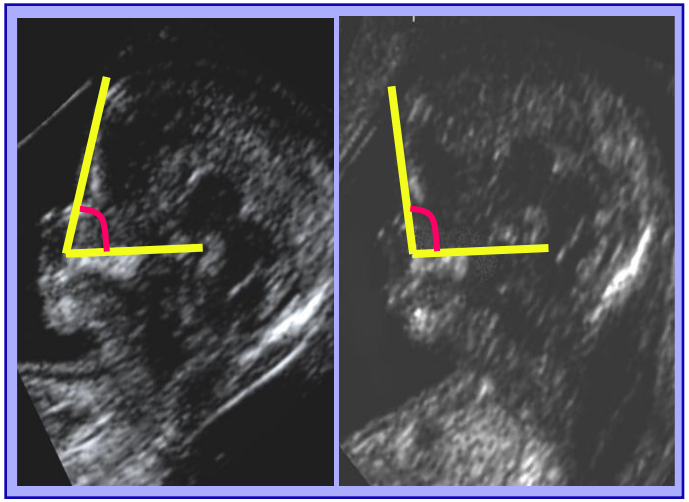
Diğer Bulgular

	Trizomi 21	Normal
NK yokluğu	**% 65**	% 2
Anormal duktus	**% 65**	% 6
Trikuspid regurjitasyonu	**% 65**	% 6
Geniş fasial açı	**% 70**	% 5









Fetal Nasal Bone Assessment
In The First Trimester Down Syndrome Screening
Has R, Kalelioglu I, Yuksel A, Ibrahimoglu L, Ermis H, Yildirim A.
Fetal Diagn Ther 2008;24:61-66

- Nazal kemik yokluğu
  - Normal: 7/1798 (%0.39%)
  - Down: 3/9 (%33.3)
    - +LHR: 85.6 (95% CI: 26.2-279.5)
    - -LHR: 0.67 (95% CI: 0.42-1.06)

	DR:	FPR:	FPR: (+NK)
Yaş+NT:	% 77.8 (7/9)	%4.5	%3.4
+(PAPP-A ve serbest β-hCG)	%88.9 (8/9)	%3.6	%3.0

First-trimester screening for trisomies 21, 18 and 13 by ultrasound and biochemical testing.

Wright D, Syngelaki A, Bradbury I, Akolekar R, Nicolaides KH.
Fetal Diagn Ther. 2014;35(2):118-26.

Kombinasyon:

- USG: NT, FHR, **DV PIV**
- Biyokimya: β-hCG, PAPP-A, **PLGF ve AFP**

METOD:

- "Model-based estimates of screening performance"

Cutoff :1:100

- **DR:**
  - **Trizomi 21 %93.3**
  - **Trizomi 18 ve 13 %95.4**
- **FPR: %1.3**

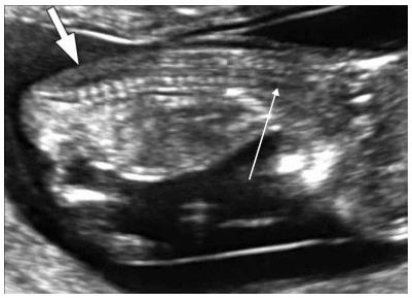
11-14 hf USG

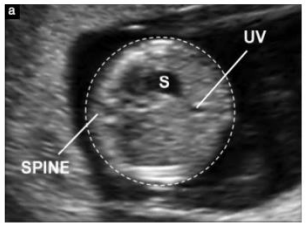
Ultrasound Obstet Gynecol 2013; **41**: 102–113
Published online in Wiley Online Library (wileyonlinelibrary.com). **DOI:** 10.1002/uog.12342

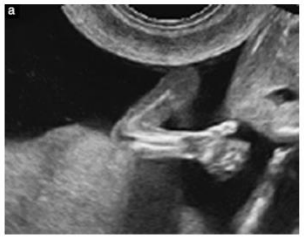
isuog.org

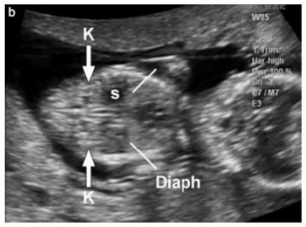
GUIDELINES

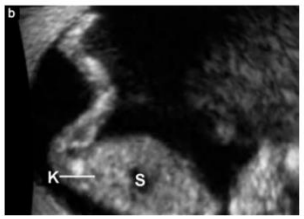
ISUOG Practice Guidelines: performance of first-trimester fetal ultrasound scan

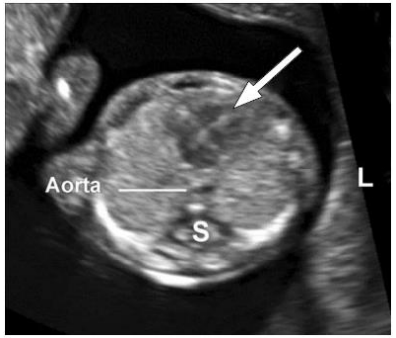


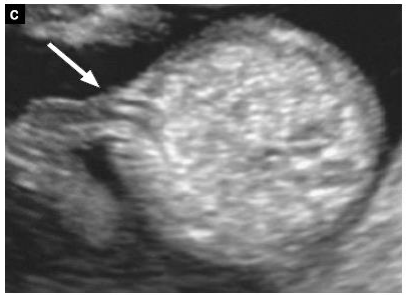


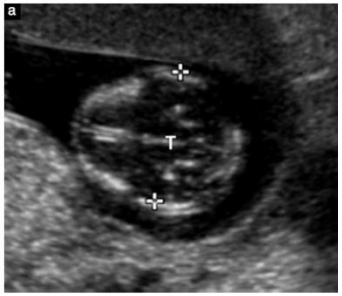


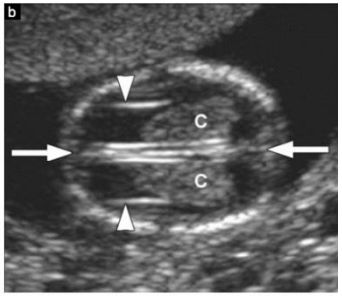


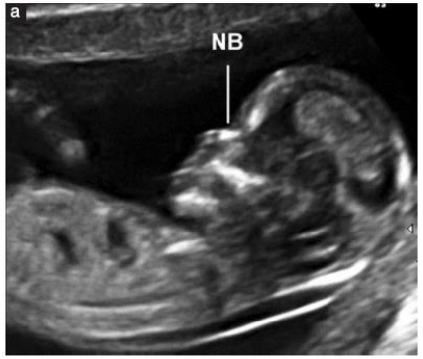


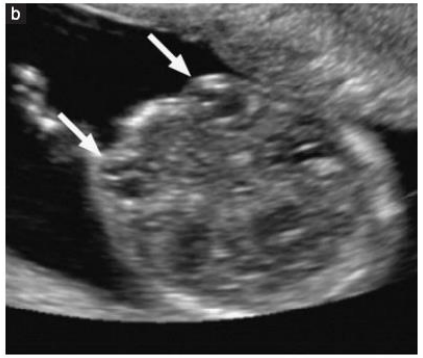


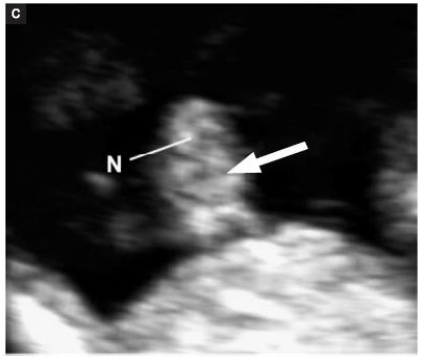




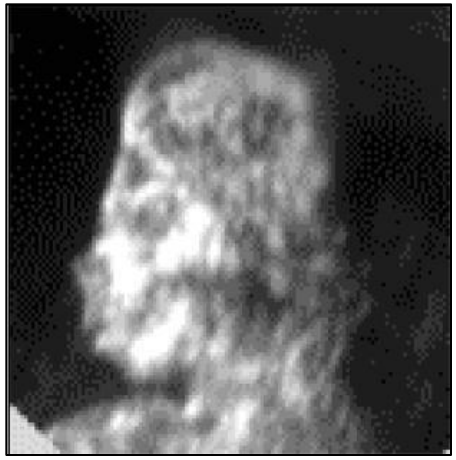




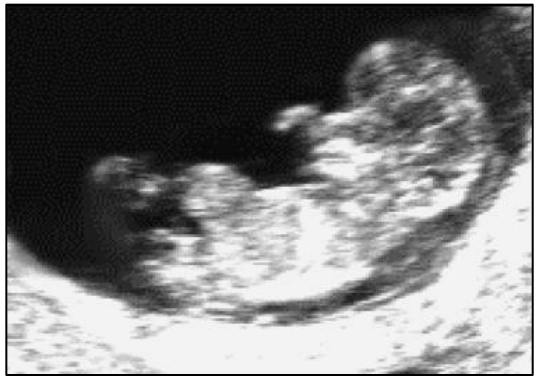


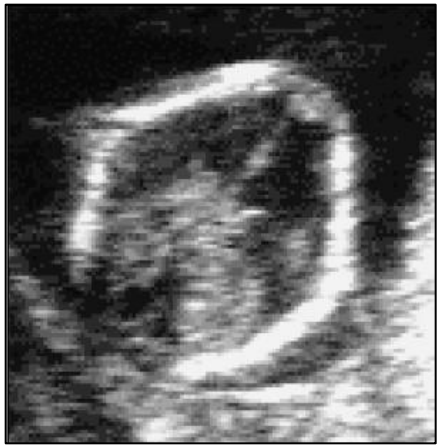


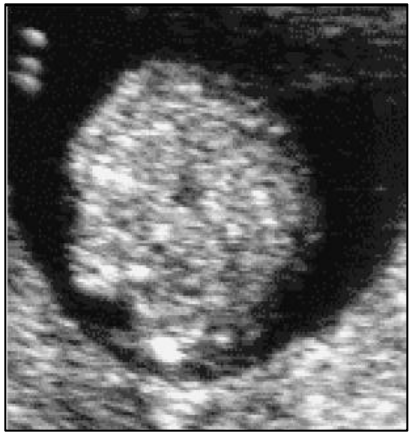
11-14 haftada fetal anomaliler

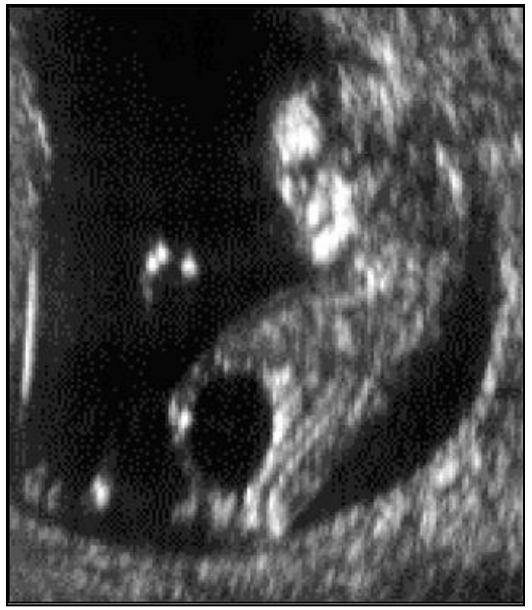


%50

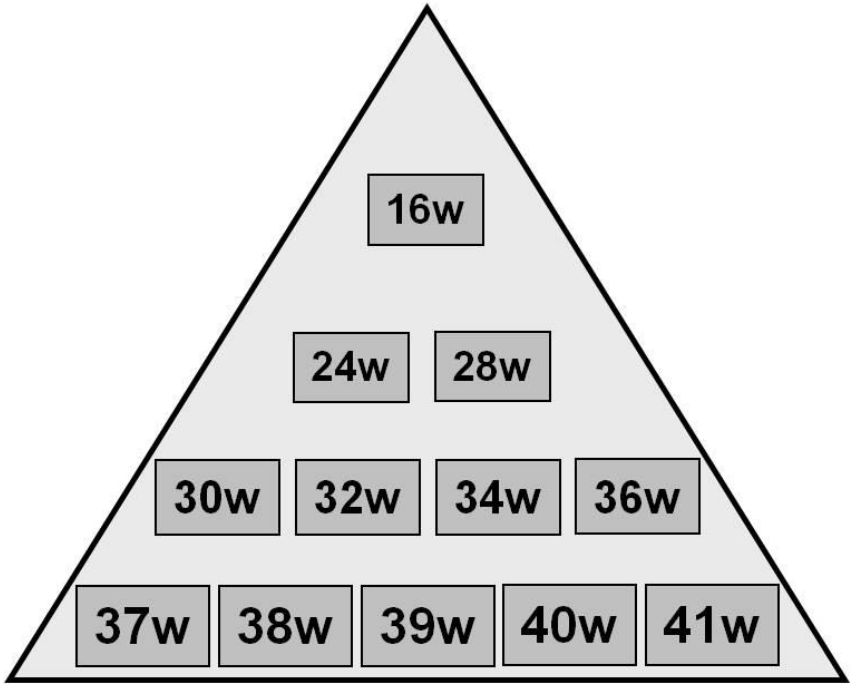


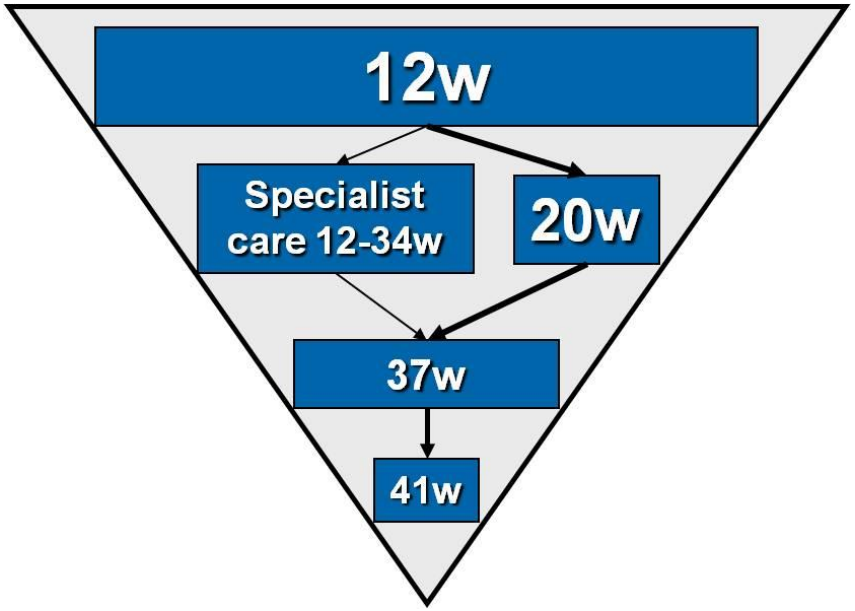






Antenatal bakım





https://fetalmedicine.com

Risk calculator for preeclampsia

The data were derived from the study of 33,602 singleton pregnancies with a live fetus at 11^{+0} - 13^{+6} weeks with no major defects or chromosomal abnormalities and delivery after 23 weeks.

Objective

- The incidence of adverse fetal and maternal short-term and long-term consequences of preeclampsia (PE) are inversely related to the gestational age at onset of the disease being worse for early-PE, requiring delivery before 34 weeks, than intermediate-PE with delivery at 34-37 weeks or late-PE delivering after 37 weeks.
- The risk for developing PE increases with maternal weight, decreases with maternal height, it is higher in women of African and South Asian racial origin compared to Caucasian women, in those with a medical history of chronic hypertension, those whose mother had developed PE and in pregnancies conceived after use of ovulation induction drugs or IVF. The risk is increased in parous women who developed PE in a previous pregnancy and lower in those who had normal pregnancies. The risk is also related to uterine artery pulsatility index (PI), mean arterial pressure (MAP) and serum pregnancy associated plasma protein-A (PAPP-A and placental growth factor (PLGF).
- It is strongly recommended that those undertaking assessment of risk receive the Fetal Medicine Foundation Certificate of competence in the 11-13 weeks scan and in Doppler ultrasound (see FMF Certificates of Competence)

Please record the following information and then press calculate.

Maternal weight	76	kg	
Maternal height	161	cm	
Racial origin	Caucasian		
	☐ Chronic hypertension		
	☐ Family history of preeclampsia		
Method of conception	Spontaneous		
Obstetric history	Nulliparous, no previous pregnancies after 23 weeks		
Fetal crown-rump length	69	mm	
Nuchal translucency	2	mm	
Uterine artery PI	1,24	MoM	The uterine artery PI MoM is derived from your measurements by the FMF calculator
Mean arterial pressure	100	MoM	The mean arterial pressure MoM is derived from your measurements by the FMF calculator
Maternal serum PAPP-A	0,96	MoM	
Maternal serum PlGF		MoM	PlGF MoM is derived from your measurements by the FMF calculator

Calculate

Back Reset the form

Birinci trimester taraması

- Kombine test: Anne yaşı+NT+sBhCG+PAPP-A
  - NIPT ile KOMBİNASYON
- “11-14 hafta ayrıntılı fetal ultrason incelemesi”
- Riskleri değerlendirme fırsatı