



FONDAZIONE IRCCS CA' GRANDA
OSPEDALE MAGGIORE POLICLINICO

Ecografia di I e II livello: dai protocolli di screening agli esami diagnostici

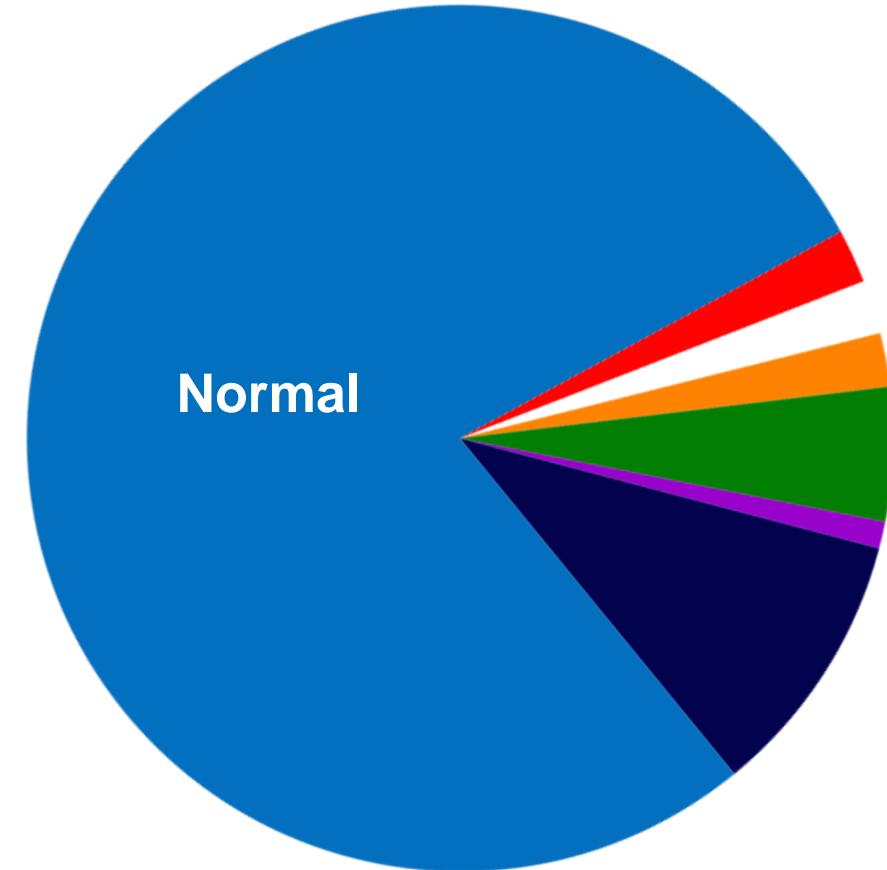
Simona Boito

Servizio di Medicina e Chirurgia Fetale
Clinica Ostetrica ‘L. Mangiagalli’
Ospedale Maggiore Policlinico, Milano





Prenatal counselling



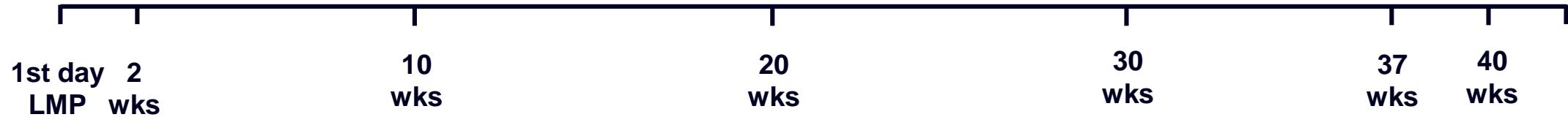
Chromosomal abnormalities	1-2%
Structural defects	1-2%
Pre-eclampsia	2%
Fetal growth restriction	5%
Early preterm delivery	1%
Others	10%



Fetal development



Embryo Fetus not viable Early preterm Late preterm Term





Prenatal Ultrasound



Timing of screening

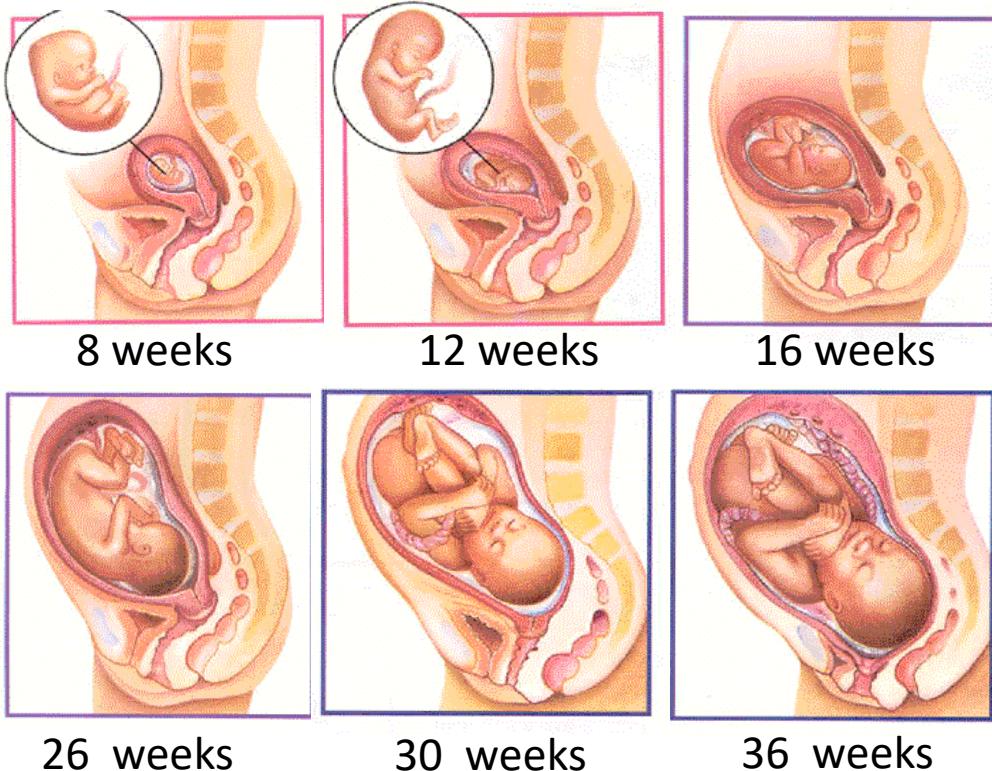
11-13 wks

19-21 wks

29-31 wks



Pregnancy dating

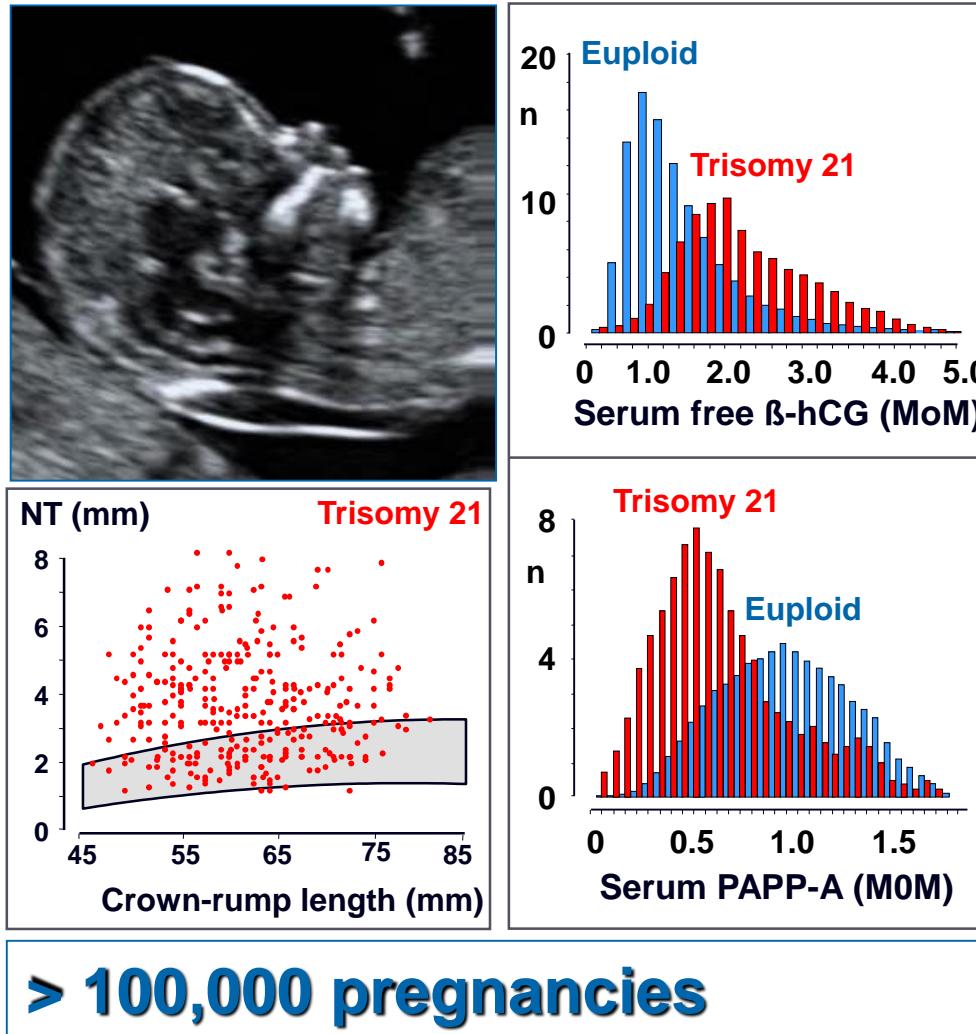


- 1st day of LMP
- Crown-rump length (CRL)
- Biparietal diameter (BPD)
- Abdominal circumference (AC)
- Femur length (FL)

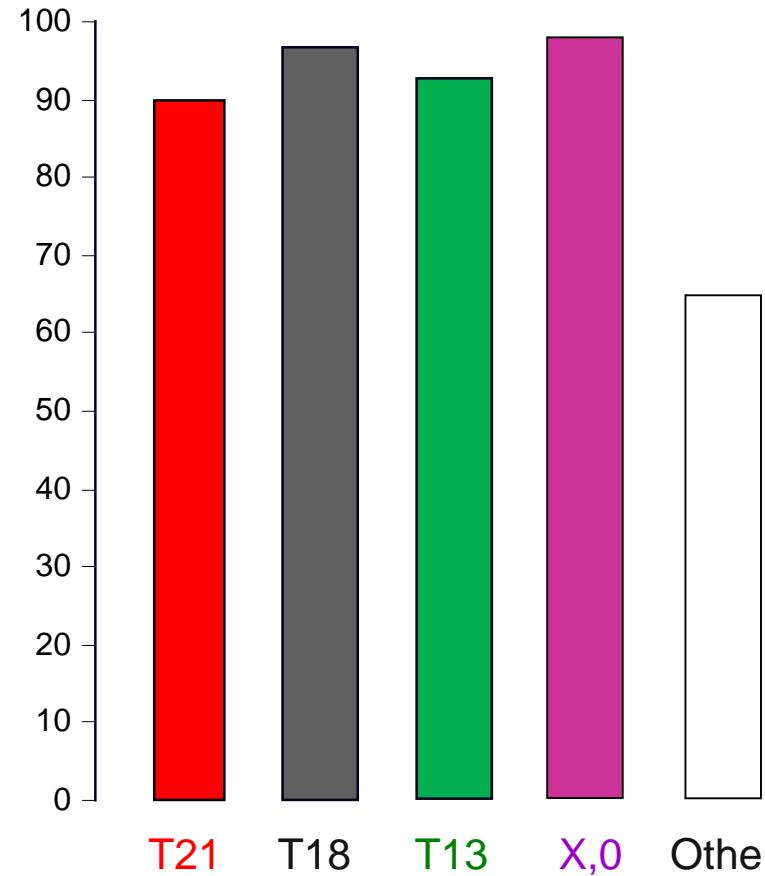


Screening for trisomy 21, 18, 13

Combined test at 11-13 weeks

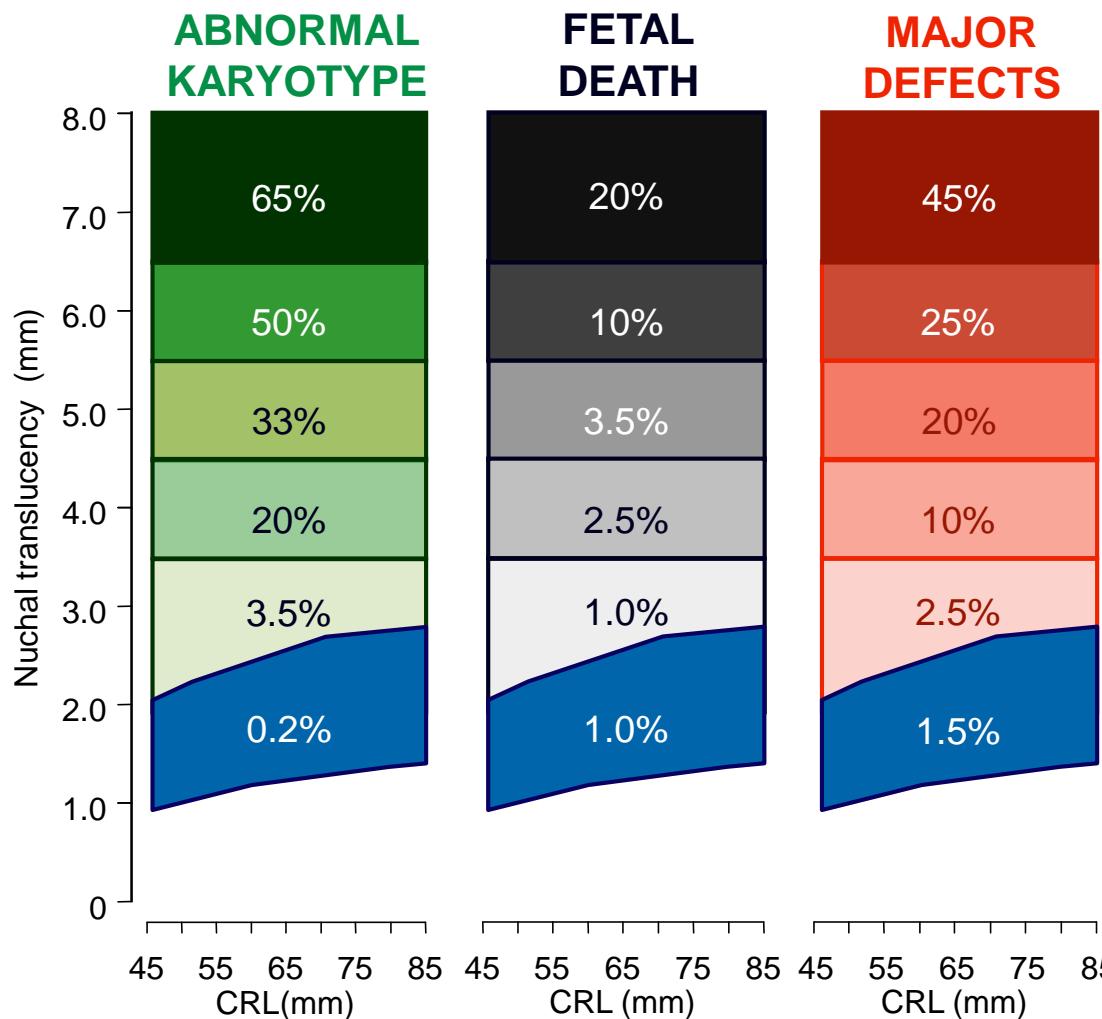


Detection rate for
FPR 4%





Implication of high NT





Early detection of fetal defects

**Ultrasound at 11-13 wks
(n=44,859)**

Major defects 1%

Detected 44%

Always detected 30%

Acrania
Holoprosencephaly
Exomphalos
Gastroschisis
Megacystis
Body stalk anomaly

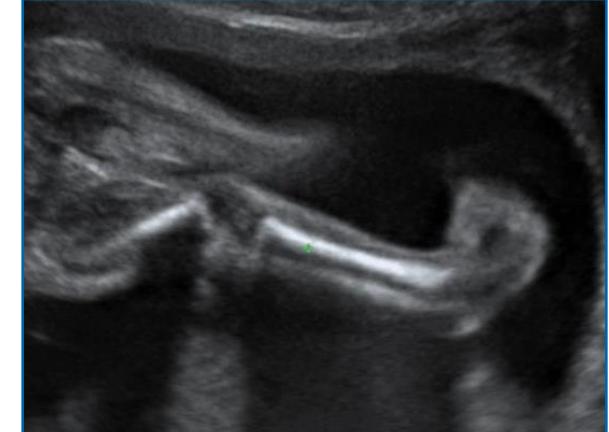
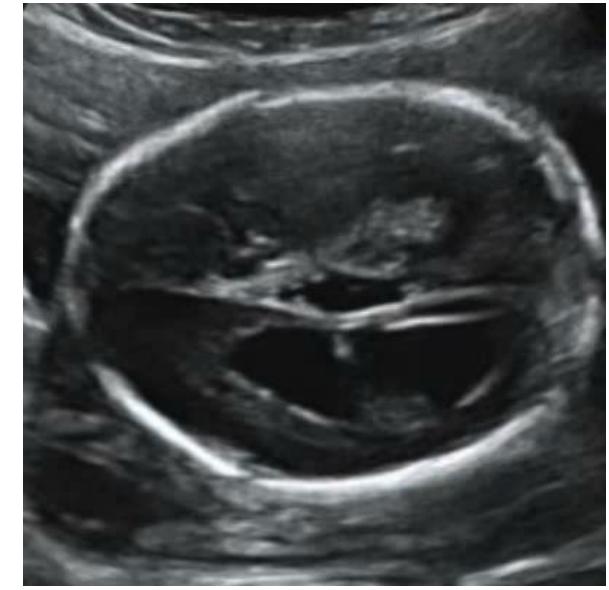




Detection of fetal defects

Undetectable at 11-13w

Corpus callosum agenesis
CCAM /sequestration
Esophageal / duodenal atresia
Bowel obstruction
Hydronephrosis
Talipes





II trimester screening

- To confirm fetal growth (and gestational age)
- To check for major structural anomalies
- To check for markers of chromosomal abnormality
- To check placental position
- To screen for preterm labour (cervical length)
- To screen for preeclampsia (uterine artery PI and MAP)



II trimester screening

Sensibilità dello screening ecografico delle malformazioni fetali

	S.N.C. %	G.E. %	Ur. %	Sch. %	Cardiov. %	Sensibilità media %
Bernaschek	68,3	46	73	53	30	50,0
Anderson	90	72	85	31	36	58,3
Chambers	92,1	24	88,4	25	18,4	50,9
Stoll	76,7	47,3	64,1	18,2	16,5	37,8
Grandjean	88,3	53,7	88,5	36,6	38,8§	61,4
Queisser-Luft	68,6	42,3	24,1	//	5,9	30,3

Legenda:

S.N.C. Sistema Nervoso Centrale

Ur. Apparato uropoietico

Cardiov. Apparato cardiovascolare

G.E. Apparato gastroenterico

Sch. Apparato scheletrico

§ Sensibilità per le cardiopatie maggiori

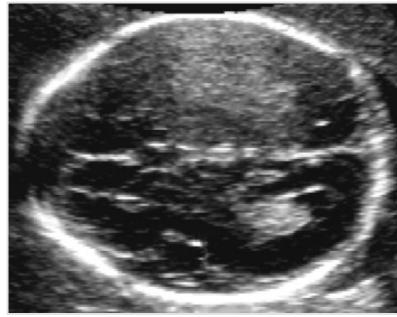
N.B. La sensibilità riportata si riferisce alle malformazioni rilevate nel secondo e terzo trimestre, in studi multicentrici.



II trimester screening

Head shape and brain

- BPD, HC view for septum cavum pellucidum and lateral ventricle
- sub-occipitobregmatic view for cerebellum and cisterna magna



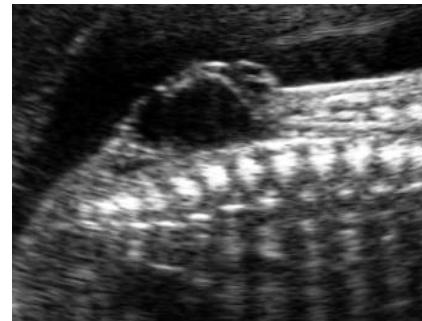
Face

- Mid-sagittal view for nasal bone and exclude micrognathia
- Transverse view of the orbits to exclude microphthalmia,
- Transverse view of the upper lip with anterior palate to exclude cleft lip



Spine

Sagittal, (coronal and transverse) views to check all vertebrae and the skin covering the spine





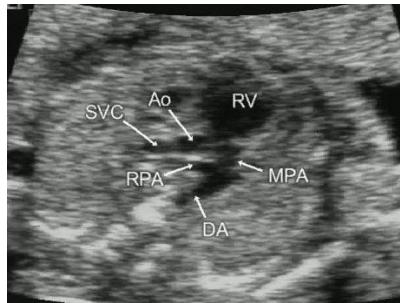
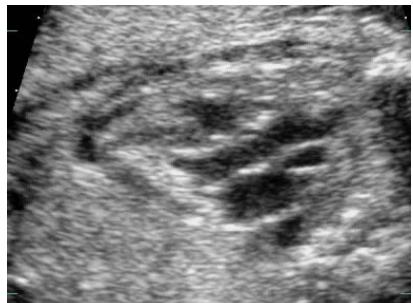
Thorax

Transverse sweep through chest and down to fetal stomach to exclude mediastinal shift and cystic lung lesions and establish situs of heart and stomach.



Heart

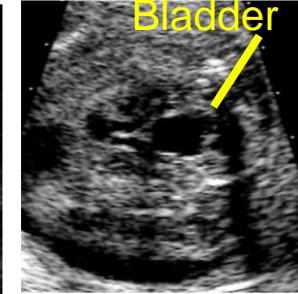
Sweep through heart in transverse plane to include 4-chamber view, outflow tracts, (3-vessel view)





Abdomen

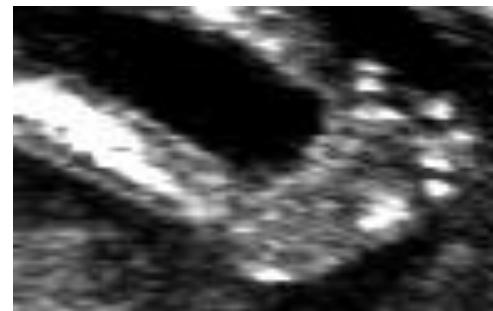
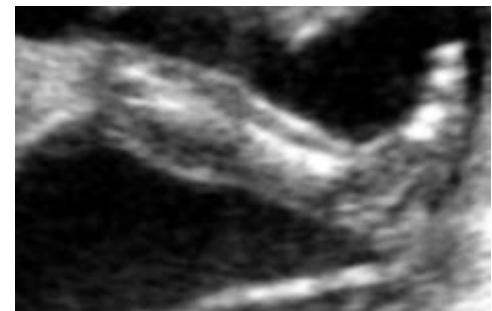
- Transverse view to demonstrate normal stomach
- Longitudinal view of the abdomen to examine: cord insertion, bladder
- Transverse view with spine up to demonstrate both kidneys and renal pelvises



Upper limbs

Lower limbs

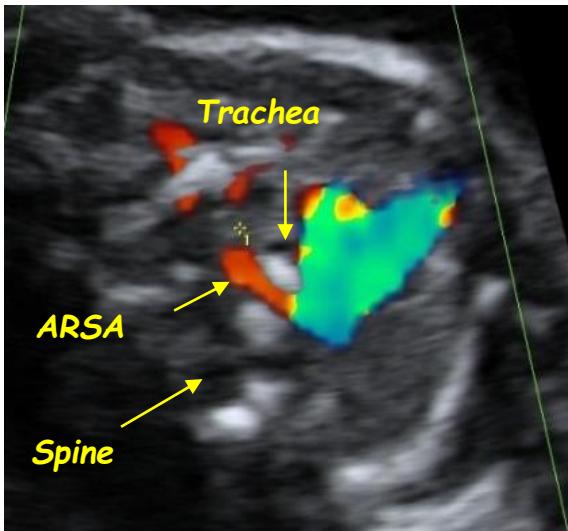
- Visualization of all long bones
- Both hands and feet
- A coronal or sagittal view of both ankles should be seen to exclude talipes. A 'sole-print' view of each foot should be obtained.



Number of umbilical cord vessels



II trimester screening





II trimester screening

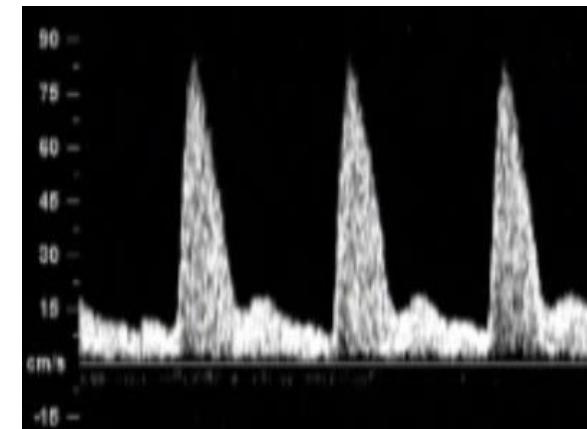
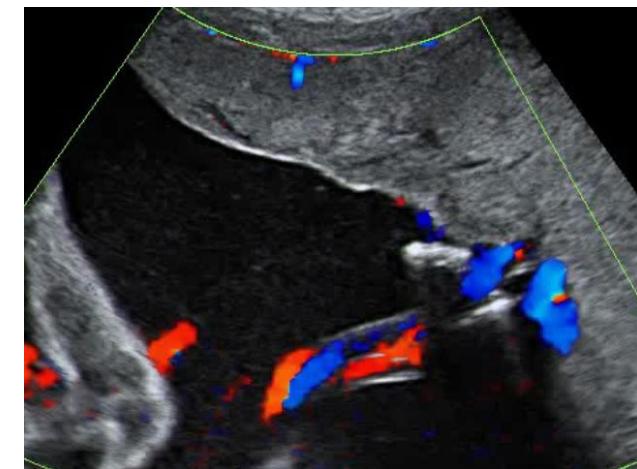
Markers of Trisomy 21

Marker	Pos LR	Neg LR
Ventriculomegaly (>10 mm)	27,52	0,94
Increased nuchal fold thickness (>6 mm)	23,30	0,80
Absent / hypoplastic nasal bone (<4,5 mm)	23,27	0,46
Aberrant subclavian artery	21,48	0,71
Hyperechogenic bowel	11,40	0,90
Bilateral hydronephrosis (>6 mm)	7,63	0,92
Intracardiac echogenic focus	5,83	0,80
Short femur (<5th centile)	3,72	0,80

- **Ventriculomegaly**
- ↑ **6-7 fold increase**
- **Nucal thickness > 6 mm**
- **Absent/hypo nasal bone**
- **ARSA**
- ↑ **3-4 fold increase**



- Placental position
- Amniotic fluid
- Uterine artery PI
- Cervical length
- Corionicity in Twins





III trimester screening

- To confirm fetal growth
- To check for major structural anomalies that develop advancing gestation
- To check placental position
- To screen for preterm labour (cervical length)
- To screen for preeclampsia (uterine artery PI and MAP)



III trimester screening

**Patologie malformative fetali a più frequente comparsa
nel III trimestre e non diagnosticabili durante
l'esame di screening del II trimestre**

Sistema Nervoso Centrale

Microcefalia

Ventricolomegalia ad insorgenza tardiva

Ipoplasia del corpo calloso

Cuore torace

Idrotorace

Stenosi valvolari aortiche e polmonari

Coartazione aortica

Forme ad esordio tardivo di cuore sinistro ipoplasico e sue varianti

Ernie diaframmatiche con erniazione tardiva dei visceri addominali

Addome

Patologia Ostruttiva Intestinale

Idronefrosi ad insorgenza tardiva

Displasie renali ad insorgenza tardiva

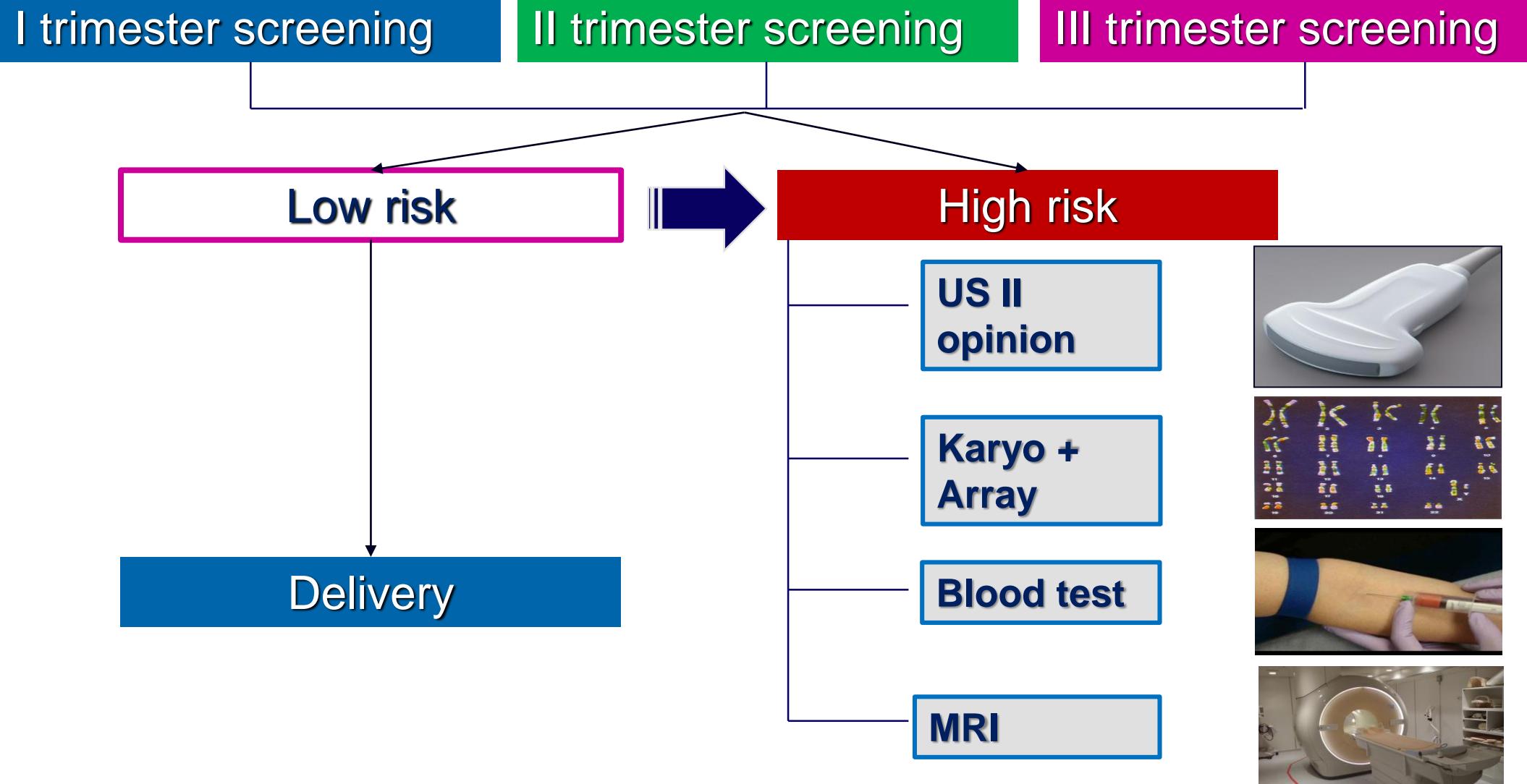
Arti

Displasie scheletriche ad insorgenza tardiva

Alcune forme di piede torto



From screening to diagnosis





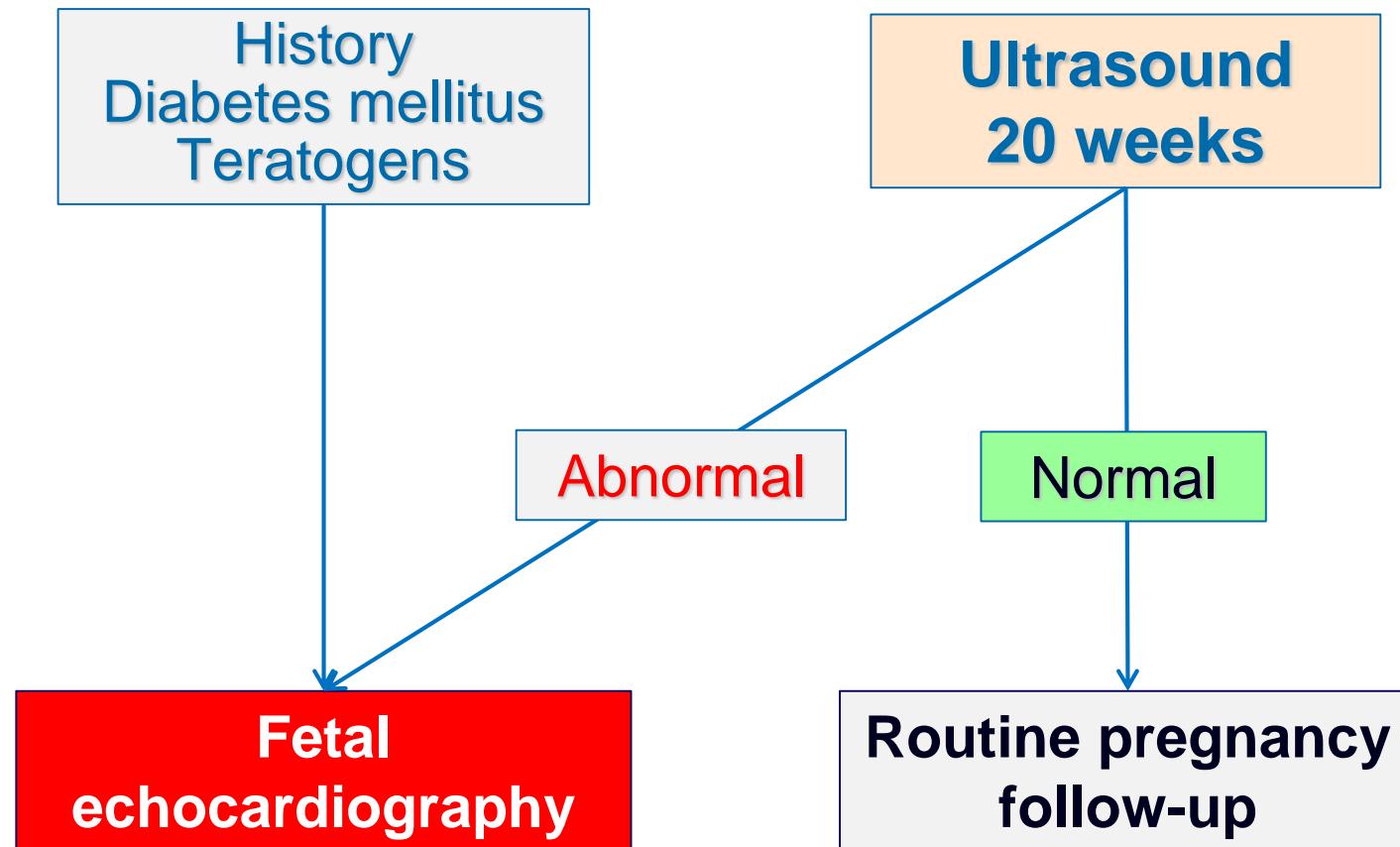
Second opinion ultrasound

“ II livello”

- Anomaly at screening ultrasound
- NT > 3 mm
- Previous anomaly
- Some drugs (not all!!!)
- Pre-conceptional diabetes
- Genetic indication
- Biometry <5° percentile
- Doppler anomalies
- Oligohydramnios 16-36 wks < 5 cm



Screening for cardiac defects in the 2nd trimester



4 chamber view



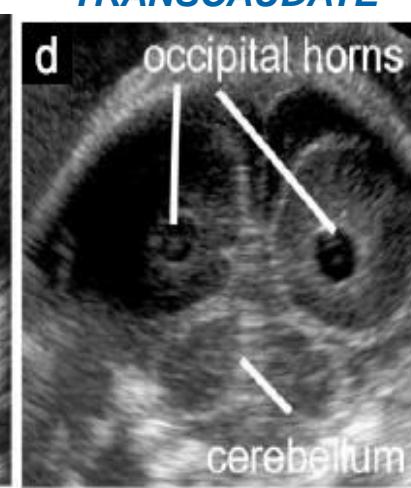
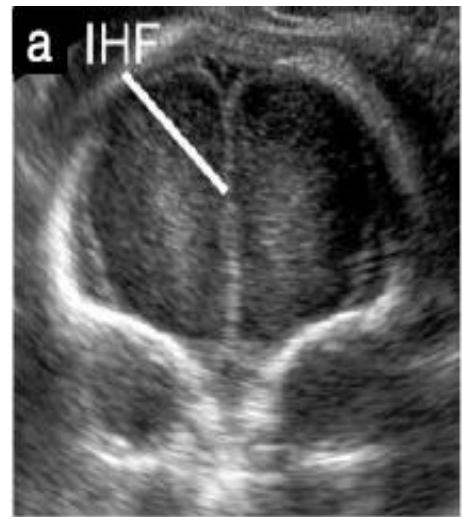
Great artery views



Detection rate 40-80%



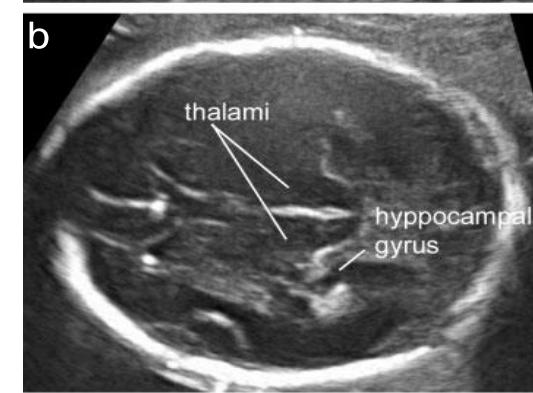
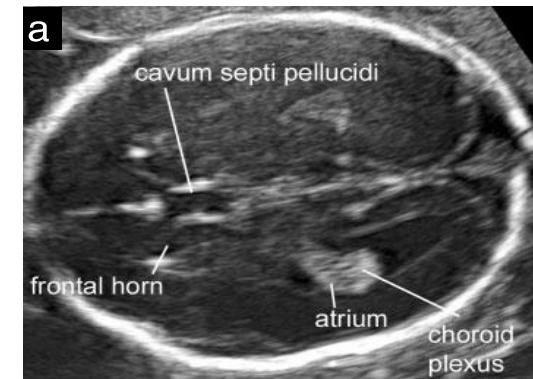
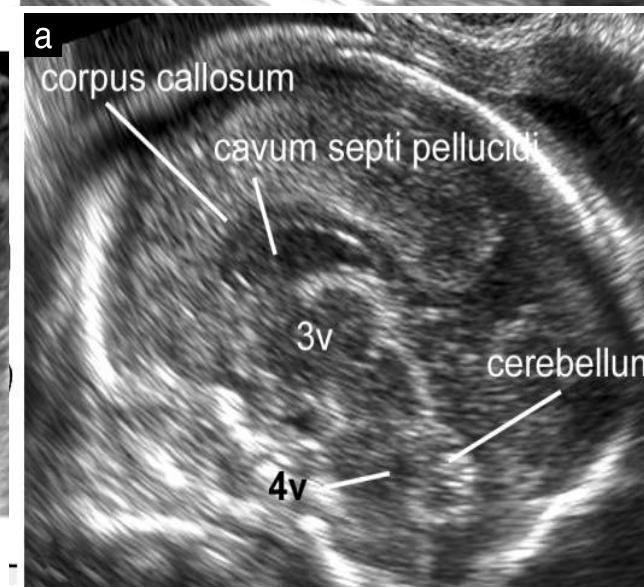
Neuros sonography vs Basic



TRANSFRONTAL

TRANSCAUDATE

TRANSTHALAMIC TRANSCEREBELLAR





Neurosonography vs Basic

Table 1 Sensitivity, specificity, positive (PPV) and negative (NPV) predictive values of ultrasound, neurosonography and magnetic resonance imaging (MRI) in predicting postnatal outcome

Method	Sensitivity (%)	Specificity (%)	PPV (%)	NPV (%)
Referral ultrasound	55	20	55	20
Neurosonography	96	87	93	93
MRI	85	80	88	75

- Normal variant without clinical implications
- Increase detection rate brain anomalies
- Decrease false positive





Invasive diagnostic testing

CVS



Amnio



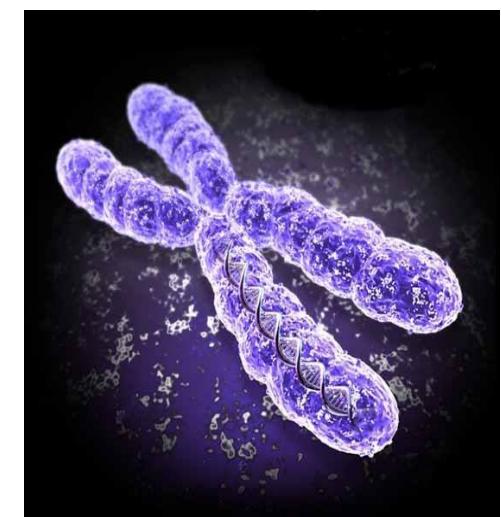
Fetal blood



Fetal karyotype



Microarray

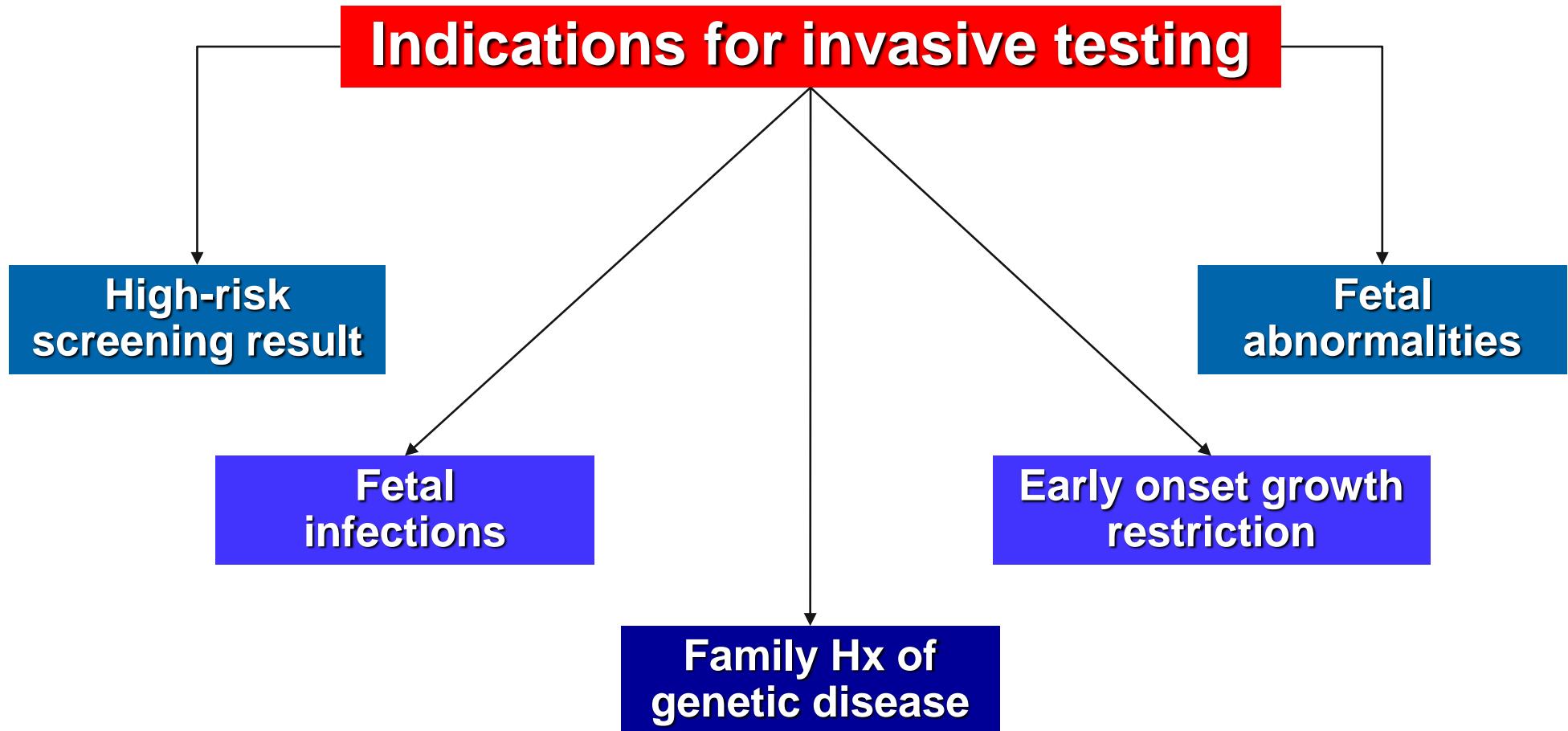


Targeted genetic testing

Haematologic testing



Indications for testing





Miscarriage from amniocentesis and CVS



?

0.2%

1.0%

2.0%



Miscarriage from amnio / CVS

National bodies - recommendations



Royal College of
Obstetricians &
Gynaecologists

Amnio 1%

CVS 1-2%



AMERICAN COLLEGE OF
OBSTETRICIANS AND
GYNECOLOGISTS

Amnio 0.3-0.5%

CVS 0.3-0.5%



THE SOCIETY OF
OBSTETRICIANS AND
GYNAECOLOGISTS
OF CANADA

Amnio 0.2-1.5%

CVS 0.2-1.5%

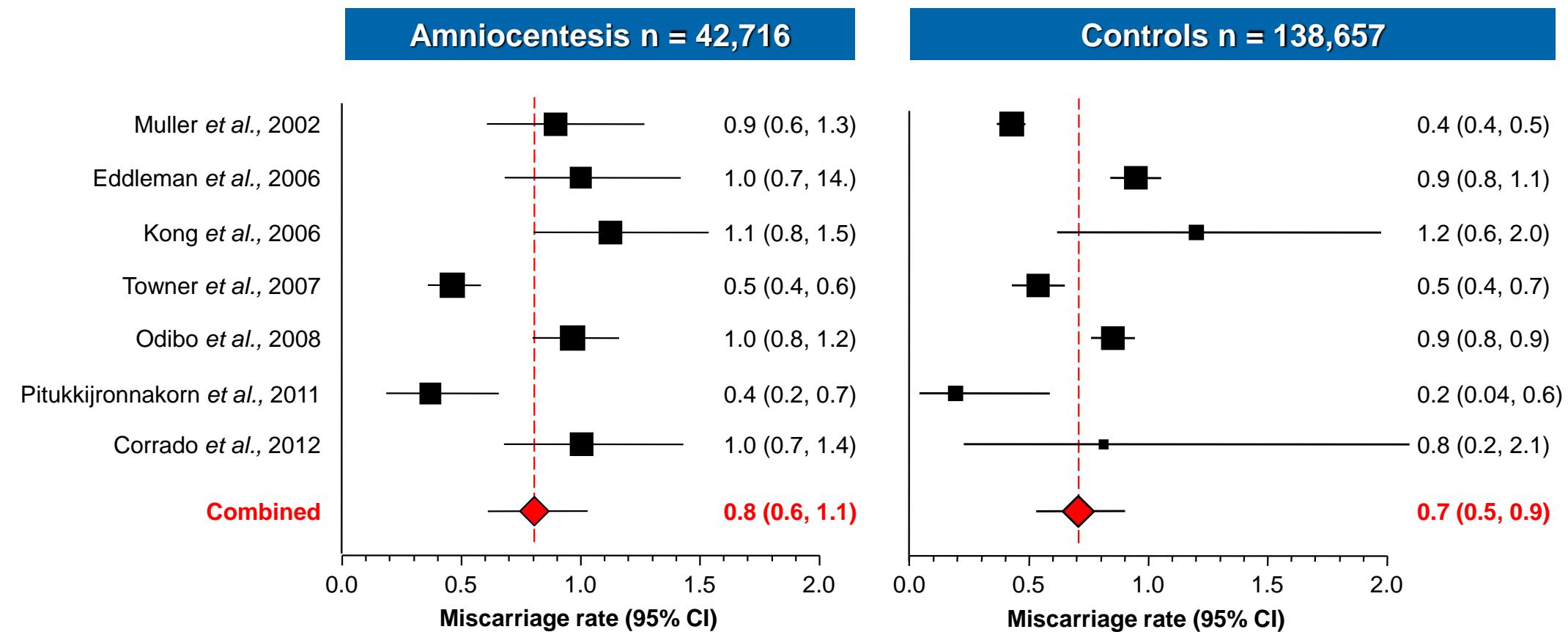
RCOG: Amniocentesis and Chorionic Villus Sampling. Green Top Guideline No.8. London: RCOG, 2010.

ACOG: Invasive prenatal testing for aneuploidy. ACOG Practice Bulletin No. 88. Obstet Gynecol 2007; 110: 1459-1467.

SOGC: Mid-trimester amniocentesis fetal loss rate. J Obstet Gynaecol Can 2007; 29: 586-595.



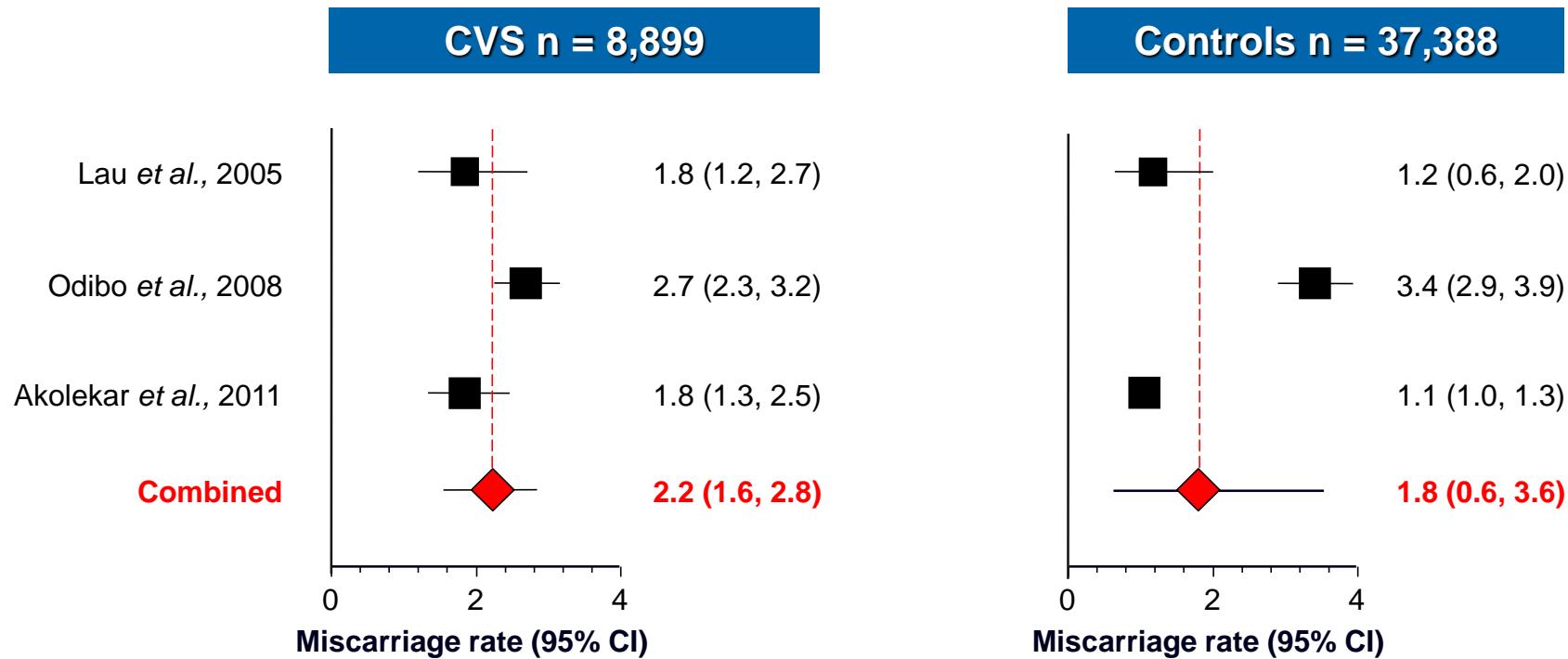
Meta-analysis – Amniocentesis studies



Amniocentesis related loss 0.11 (-0.04 to 0.26)



Meta-analysis – CVS studies



CVS related loss 0.4 (-0.7 to 1.2)



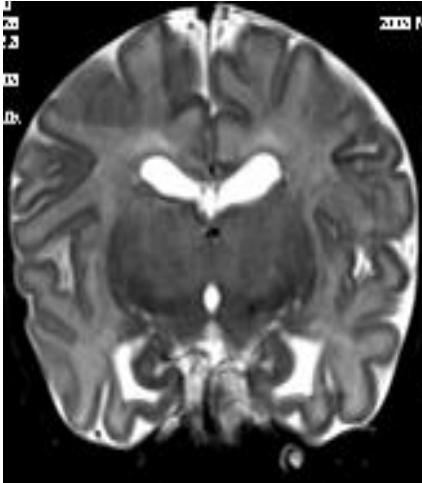
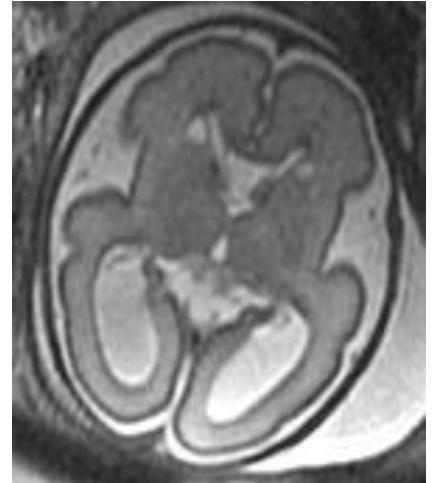
From screening to diagnosis

Blood test

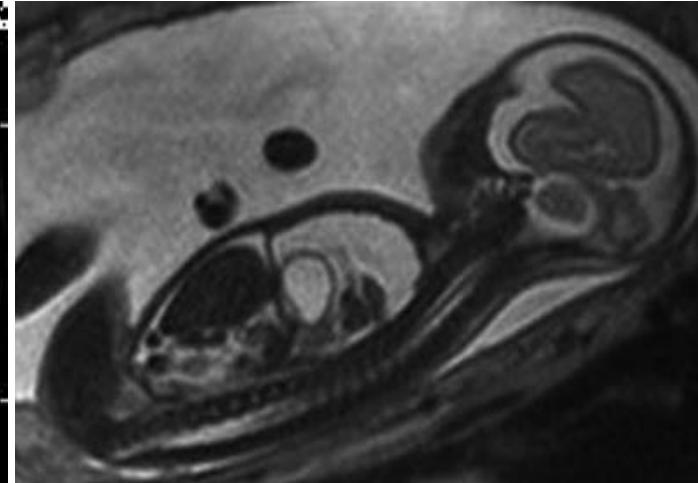
- Alloimmunization against erythrocyte antigens
- Infections
- Alloimmune thrombocytopenia

MRI

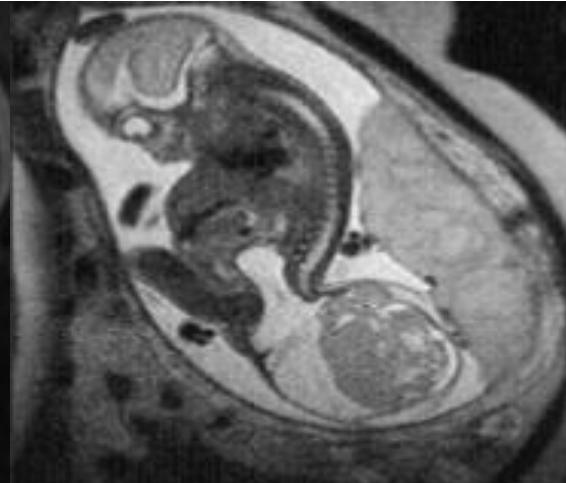
Brain



Torax



Abdomen





Fetal surgery

When minimally invasive fetal surgery is an option

- Accurate diagnosis and staging possible
- Natural history of the disease is documented
- Prognosis is established
- Intervention performed in specialized centers

Complicated monochorionic multiple pregnancies

Diaphragmatic hernia

Cystic Pulmonary Adenomatoid Malformation (CPAM)

Pulmonary sequestration

Pleural effusion / Hydrothorax

Lower urinary tract obstruction

Fetal and placental tumors

Spina Bifida





14:34

 Chirurgia Fetale

 Chi Siamo

 Come Funziona

 Dati Registrazione

 Richiedi Consulenza

 Contattaci

 Come Raggiungerci

 News

14:34

 Dati SALVA

Medico/Paziente	Medico
Nome	Nicola
Cognome	Persico
Ospedale/Centro	Opzionale
Città	Milano
Telefono	Opzionale
Email	nicola.persico@gmail.com

14:35

 Caso Clinico INVIA

Iniziali Paziente	IC
Data di Nascita Paziente	30/09/1976
Numero Feti	2
Corialità	Monocoriale
Amnioticità	Biamniotica
Età Gestazionale	20sett, 0gg
Indicazione	TTTS
Immagini	Opzionale
Urgenza	48h
Telefono Paziente	Opzionale

14:37

 Casi Clinici NUOVO

Inviati

Iniziali paziente: IC	30/09/2015
Età: 39	
Epoca Gestazionale: 20sett, Ogg	
Indicazione: TTTS	
Numero Feti: 2	
Urgenza: 48h	
	
	

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Thank you