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First Trimester Screening: New directions

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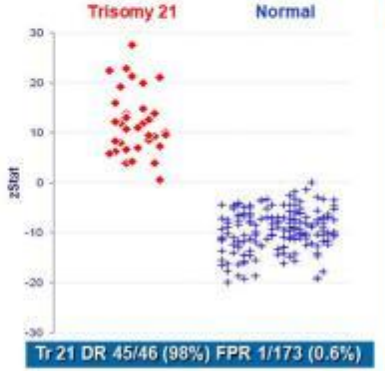
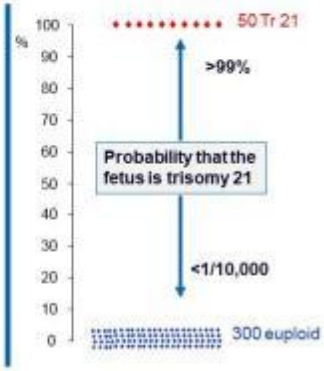
Non invasive diagnosis of trisomy 21
Selective sequencing of cell-free DNA in maternal blood





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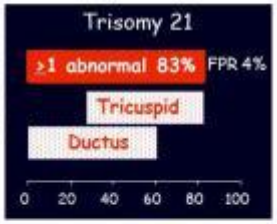
ductus venosus / tricuspid regurgitation

Trisomy 21: 65%

Normal: 3.5%

Trisomy 21: 55%

Normal: 1%



FMF screening (n=8,823)

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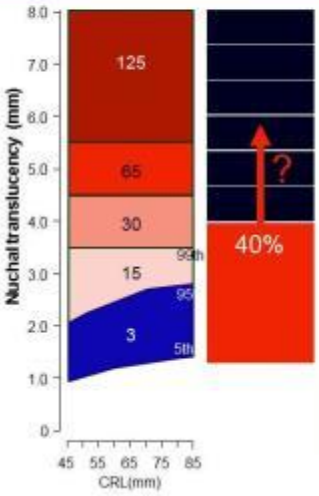
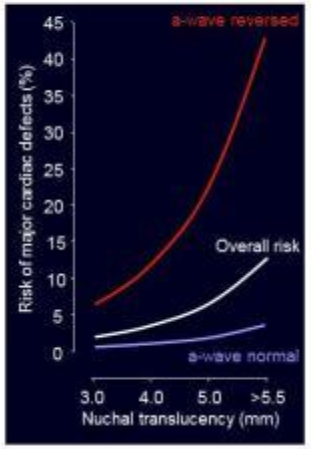
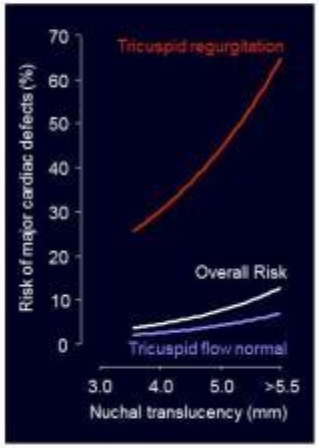
Screening for major cardiac defects







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Screening for major cardiac defects

$$\text{Fetal specific risk for major cardiac defect} = \frac{e^{(-6.924+0.833\delta NT + 2.039DV(rev) + 2.841TR)}}{(1+e^{(-6.924+0.833\delta NT + 2.039DV(rev) + 2.841TR)})}$$

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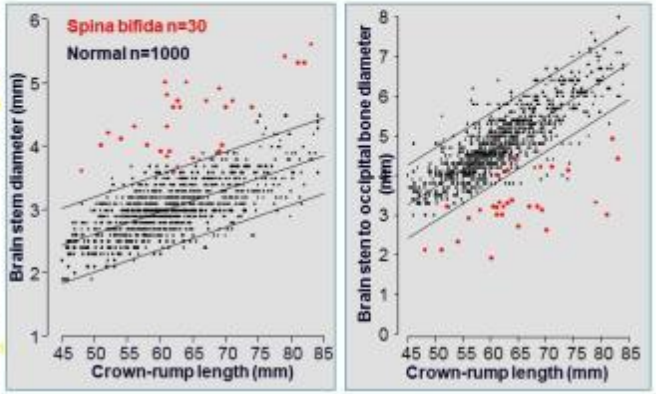
Early detection of Open spina bifida



Chaoui *et al.*, 2009; Lachman *et al.*, 2011

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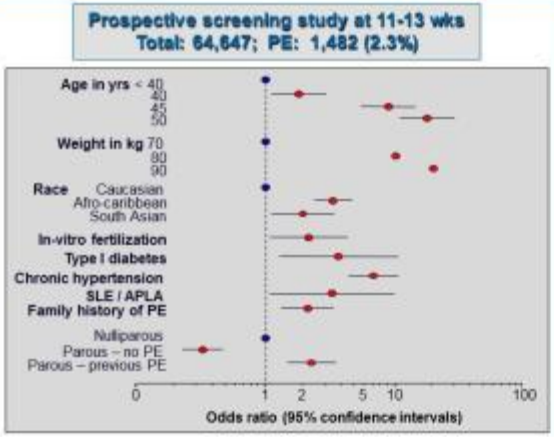
Early prediction of preeclampsia

Maternal history: *a priori risk*



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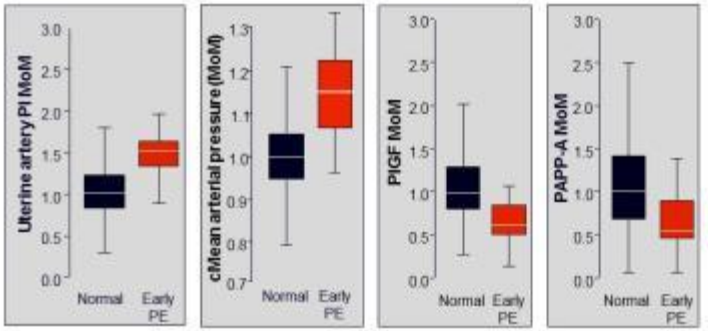
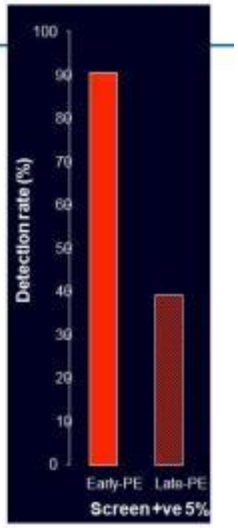
First trimester screening: PET / IUGR



Poon *et al.* 2009



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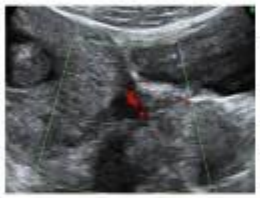
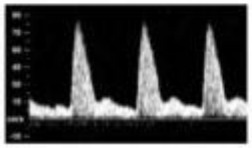
Uterine artery Doppler and aneuploidy





	n	PAPP-A (MoM)	Free β-hCG (MoM)	Uterine artery PI (MoM)
Controls	1864	1.01 (0.72-1.42)	0.97 (0.66-1.47)	1.00 (0.83-1.22)
Late PE	132	0.90 (0.56-1.24)	0.98 (0.65-1.50)	1.20 (0.95-1.46)*
Early PE	33	0.58 (0.39-1.01)*	1.12 (0.72-1.95)	1.52 (1.30-1.67)*
Trisomy 21	200	0.54 (0.38-0.82)*	2.29 (1.47-3.34)*	1.02 (0.81-1.26)
Trisomy 18	55	0.22 (0.13-0.37)*	0.27 (0.14-0.41)*	1.20 (0.96-1.35)*
Trisomy 13	19	0.32 (0.22-0.47)*	0.55 (0.39-0.91)	1.24 (1.04-1.33)
Turner	19	0.51 (0.26-0.88)*	1.19 (0.71-1.92)	1.29 (1.07-1.59)*
Triploidy	8	0.08 (0.04-0.14)*	0.18 (0.08-0.43)*	1.20 (1.02-1.40)

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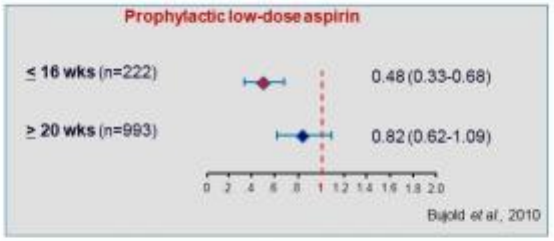
Early prediction of pre-eclampsia

Management



12 w: History, Uterine PI, MAP, PLGF / PAPP-A

- Low risk
- High risk → Aspirin

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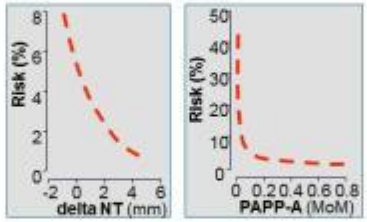
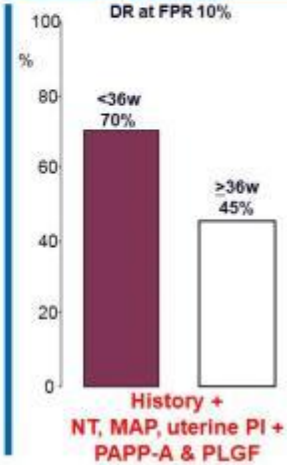
Early prediction of small babies (no PE)

History, NT, uterine PI, MAP, PAPP-A, PLGF at 11-13wks

Maternal factors	OR
Weight	0.81
Height	0.96
Race	
Caucasian	1
African	2.3
South Asian	2.4
Nullipara	1
Previous small baby	1.8
Chronic hypertension	1.9
Assisted conception	1.6
Smoking	2.6

Karagiannis *et al.*, 2011





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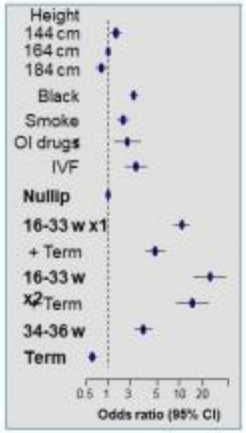
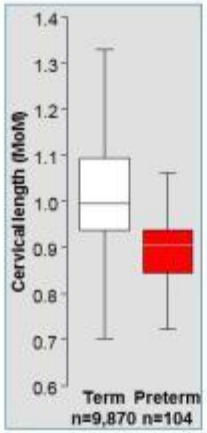
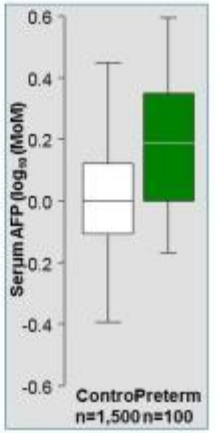
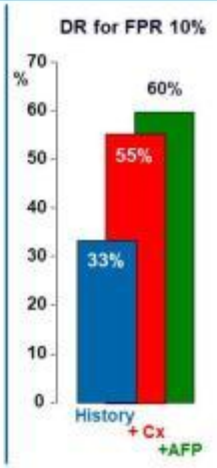
Prediction of spontaneous preterm birth

History, cervical length, serum AFP









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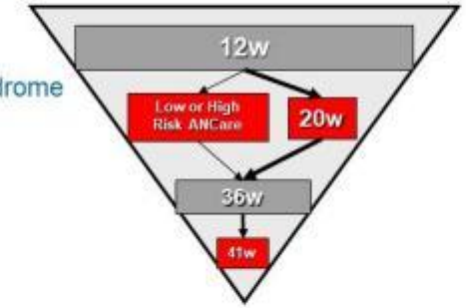
The Pyramid of Care

12 weeks predictive modeling for:

- Chromosomal abnormality
- Pre-eclampsia / IUGR / abruption
- Preterm delivery
- Major cardiac defects
- Twin twin transfusion syndrome



36 weeks predictive modeling for:

- Stillbirth / Hypoxic Ischaemic Encephalopathy
- Caesarean delivery
- Perineal trauma / shoulder dystocia