

Translucenza
ispessita
e cariotipo normale:
cosa fare ora ?

05 Giugno 2010

Milano Marittima

Silvia Malaigia

NT aumentata



-Aneuploidie

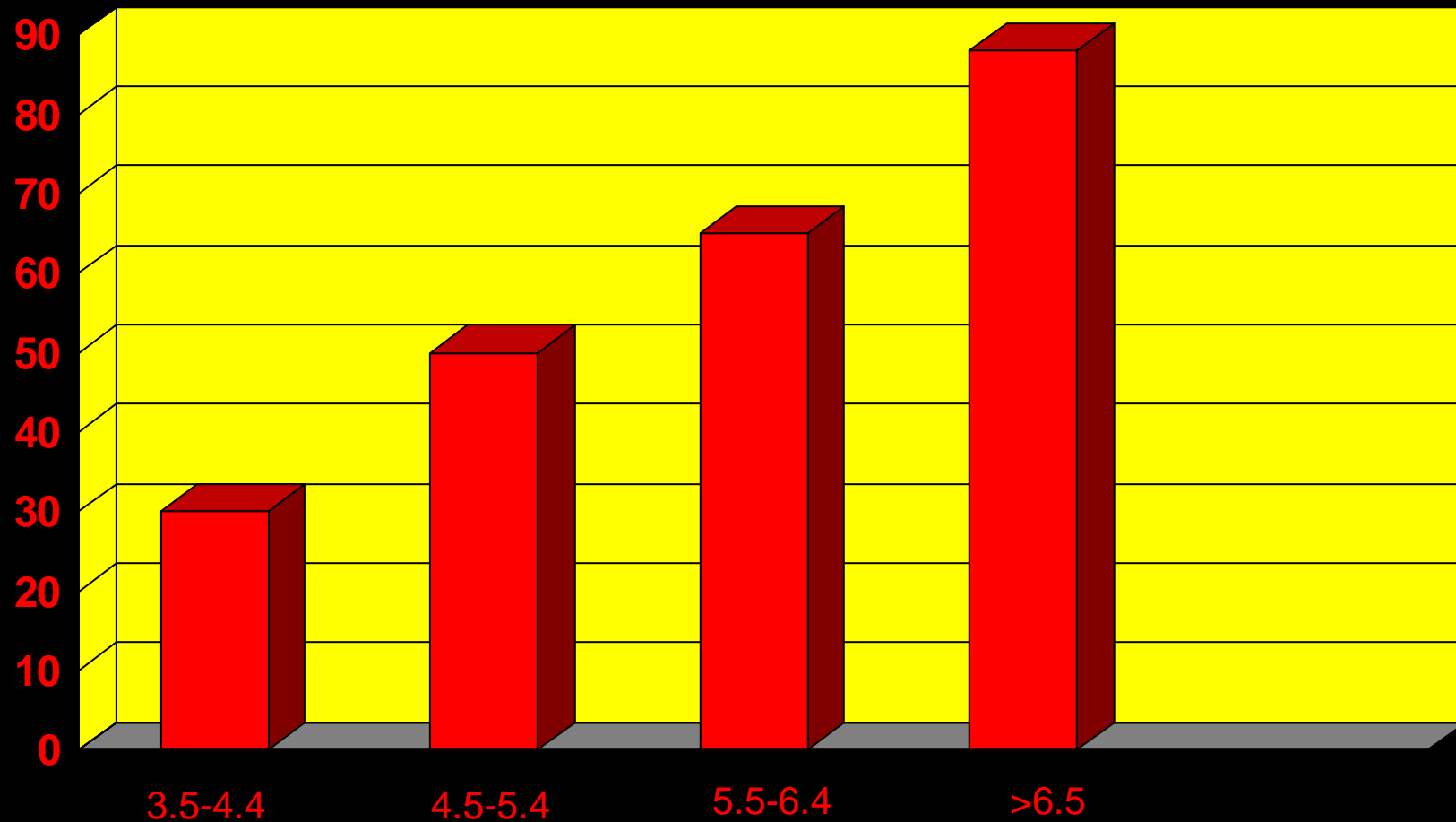
-Aborto, MEF

-Malformazioni

-Sindromi
genetiche

Incidenza di outcome sfavorevole in funzione di NT

Souka et al., 2001



NT

Aborto e MEF

-1,6% 95°<NT>99°th percentile

-20% NT > 6,5 mm

Souka et al., 2005; Bilardo et al., 2007

Anomalie strutturali

2,5% $95^\circ < NT > 99^\circ$

45% $NT > 6,5 \text{ mm}$

Anomalie strutturali

Cardiopatie

Labiopalatoschisi

Ernia diaframmatica

Displasia scheletriche

Anomalie renali

Onfalocele

cardiopatie

1% 2,5-3,4 mm

3% 3,5-4,5 mm

7% 4,5-5,4 mm

20% 5,4-6,4 mm

30% > 6,5 mm

Non c'è associazione tra valore di NT
ed una specifica cardiopatia

Sindromi genetiche

Noonan

Di George

Smith-Lemli-Opitz

Atrofia muscolare spinale

Beckwith-Wiedemann

Mecckel-Gruber

Apert

Pallister Killiam

Tar syndrome

Management della gravidanza con NT aumentata

11-13 w
Cariotipo
ecografia

14-16 w
Ecografia+
ecocardio

Eco 20 W+
ecocardio

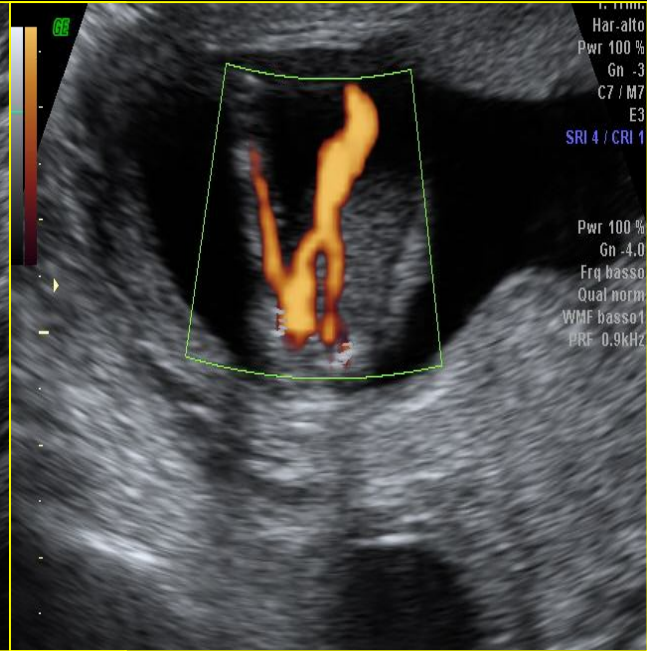
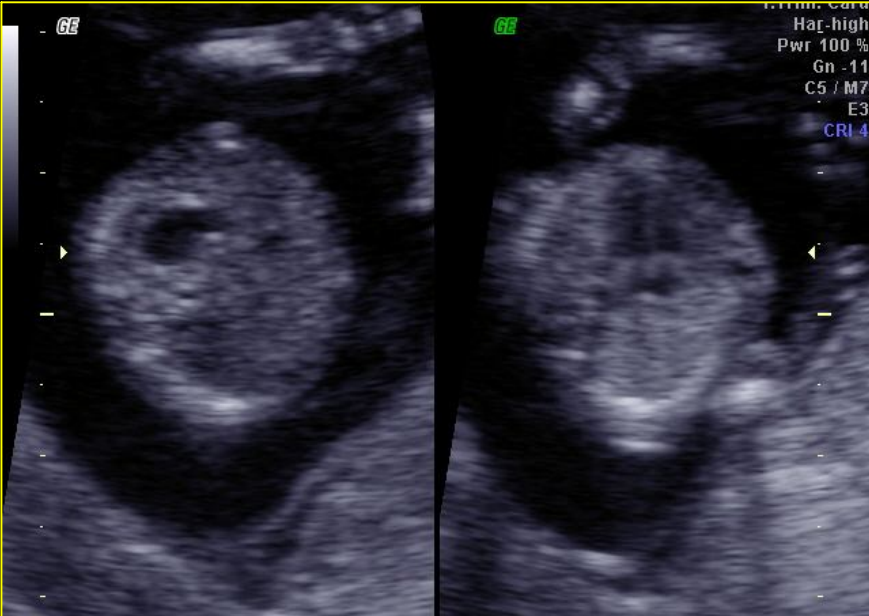
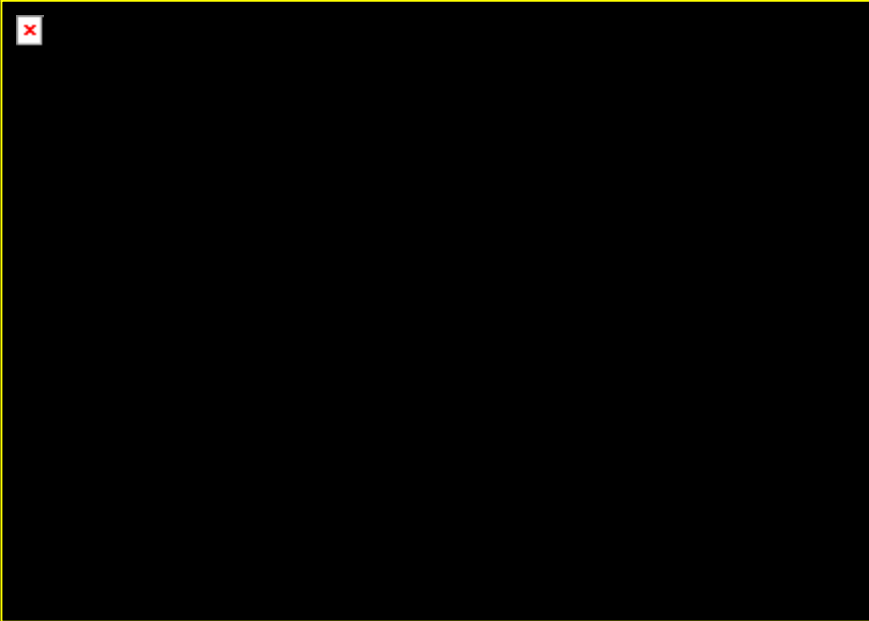
Screening
infett.
test genetici
22q11

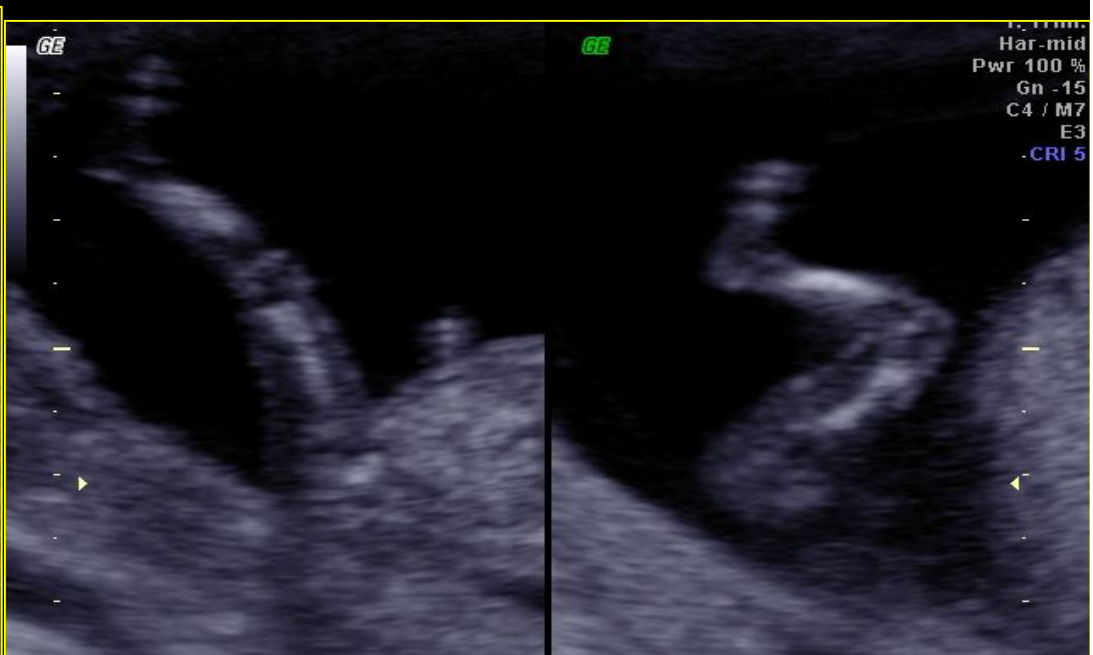
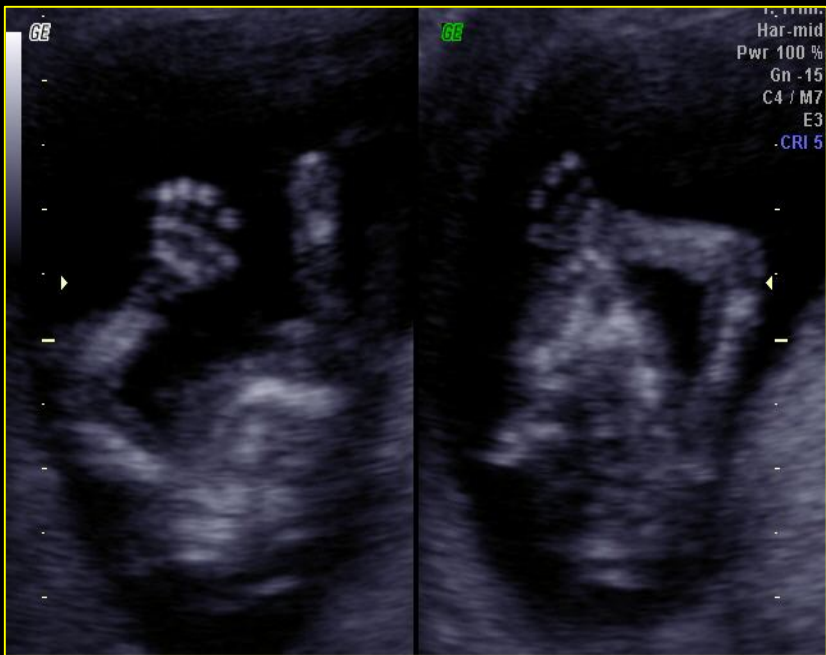
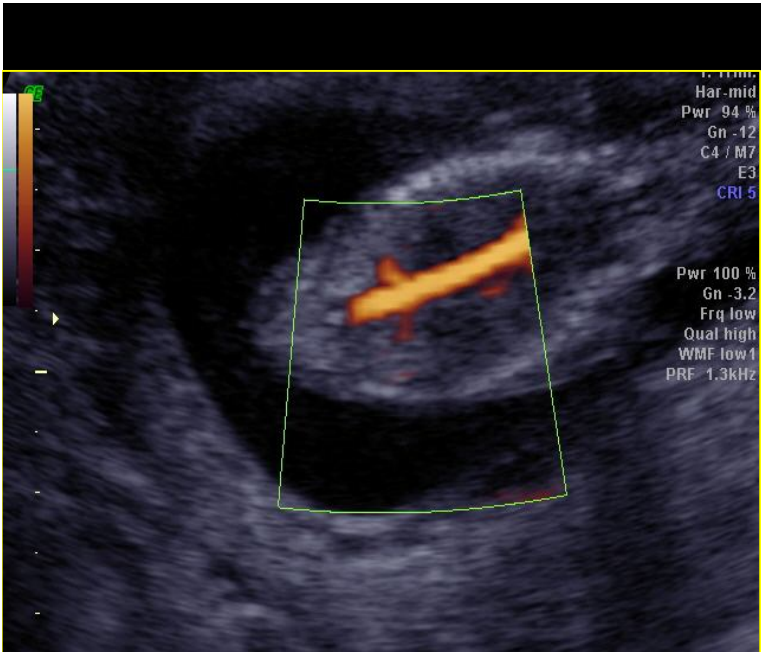
In epoca postnatale
eventuale consulenza genetica

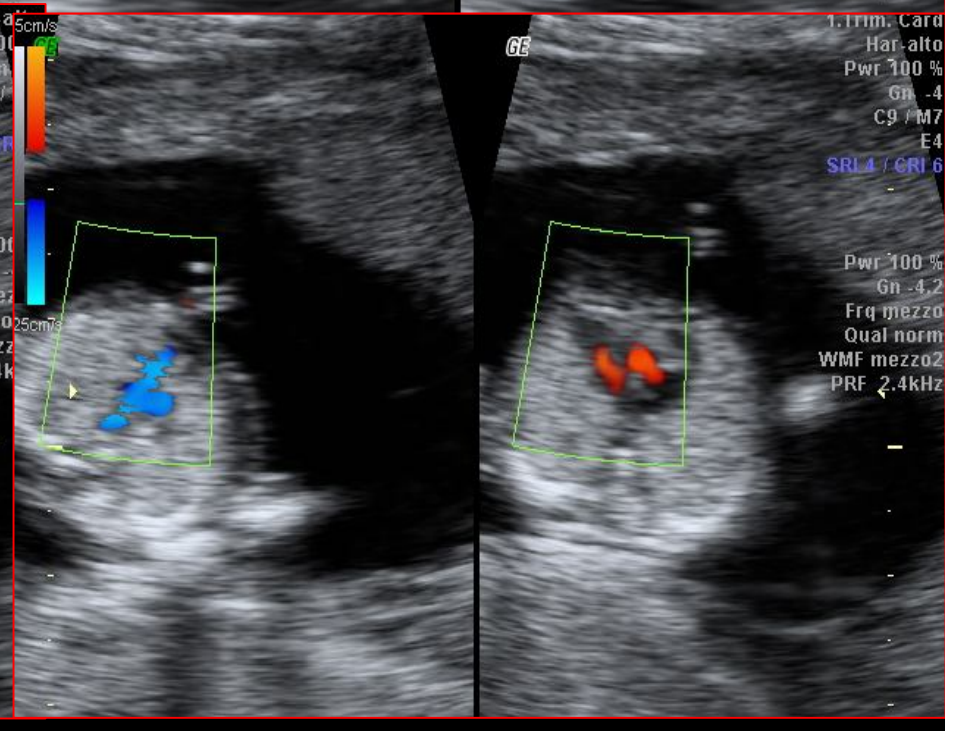
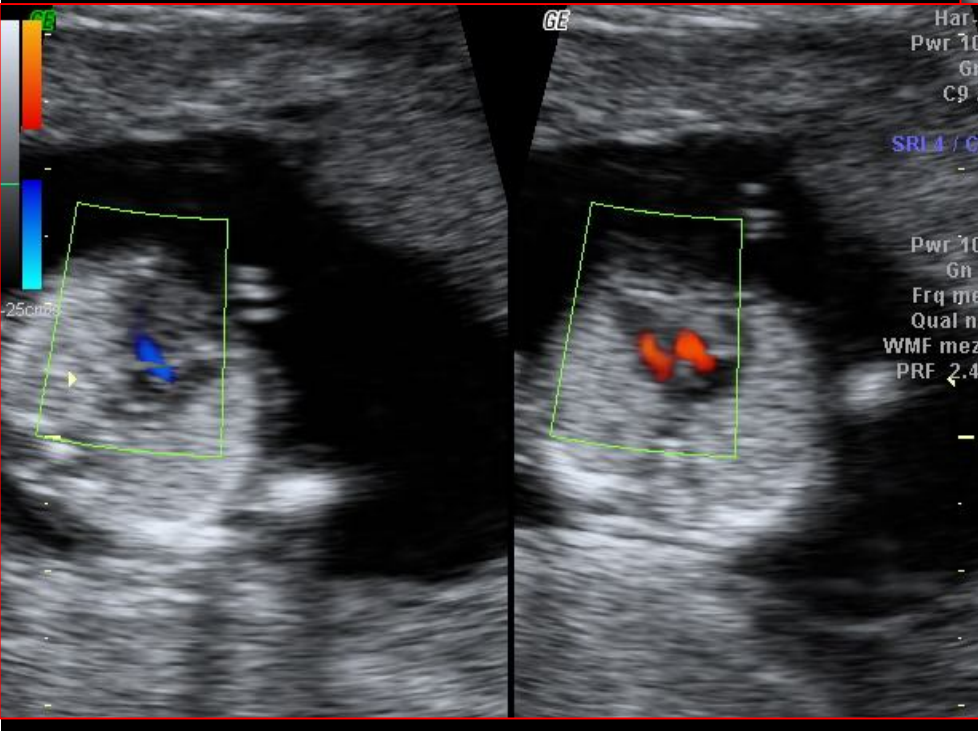
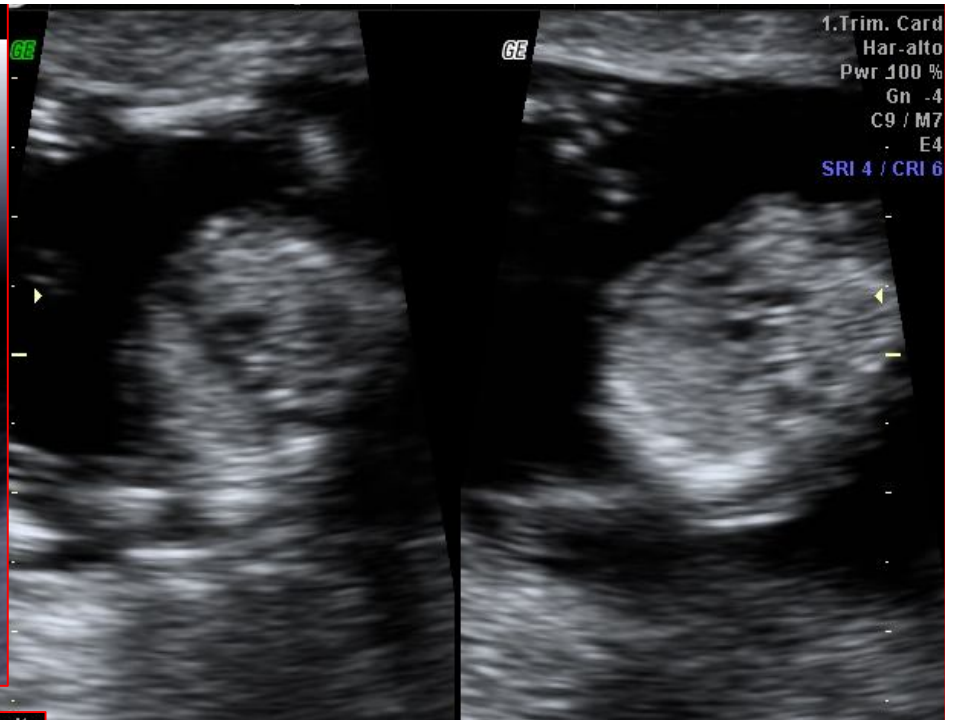
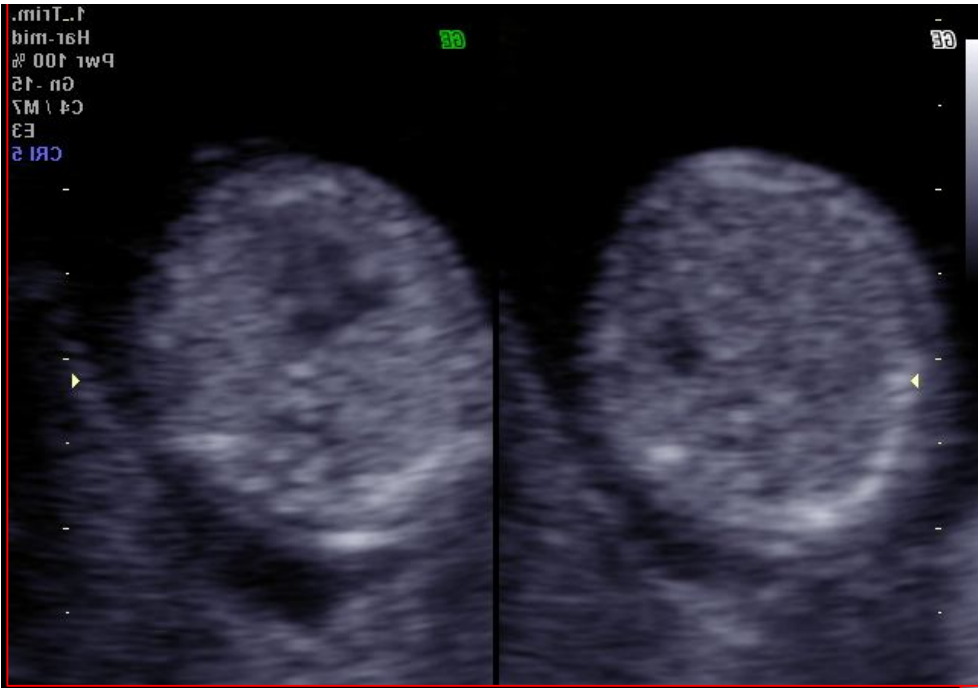
Ecografia morfologica precoce

- 75% a 11 w
- 96% a 12 w
- 98% a 13-14 w

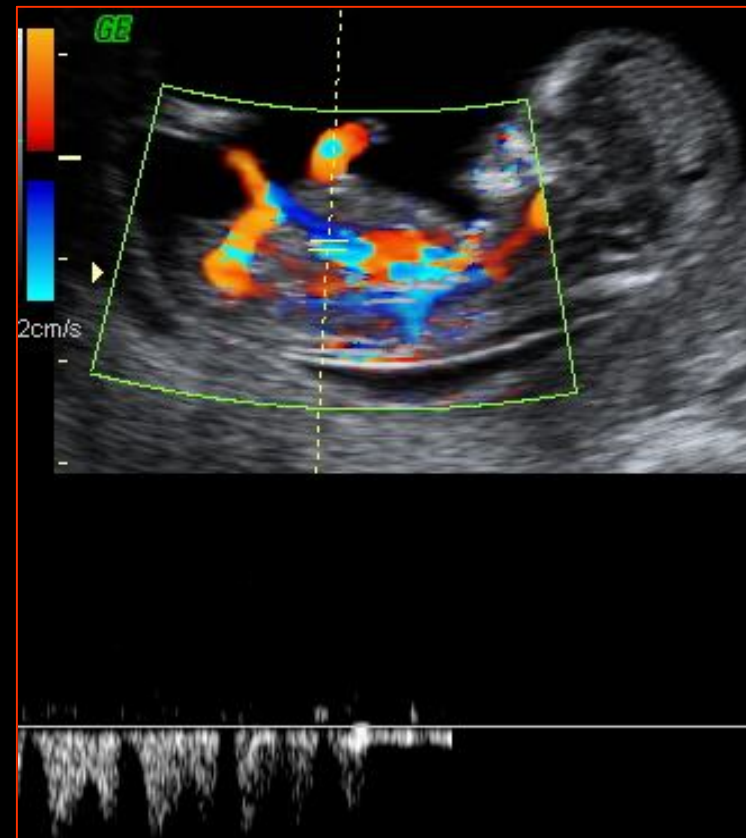
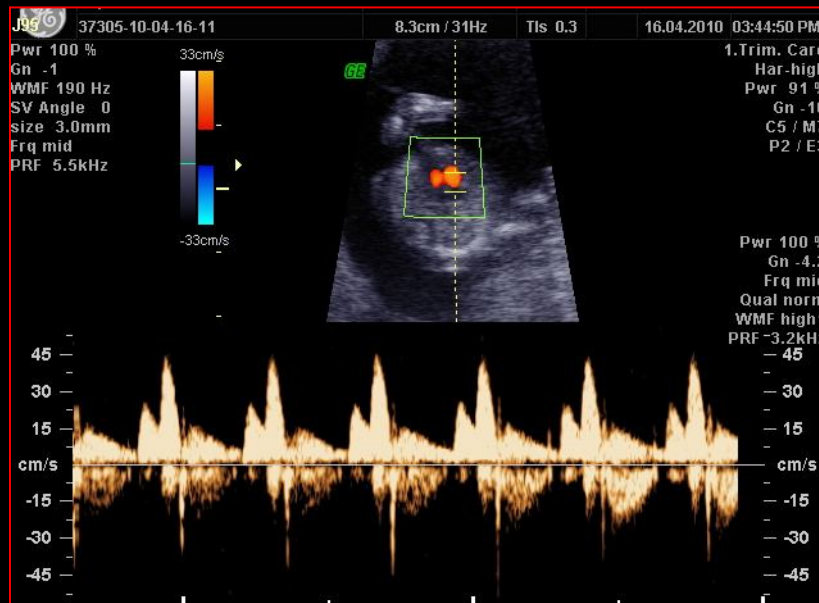
A.P. Souka Ultrasound Obstet Gynecol
2004;24:730-734



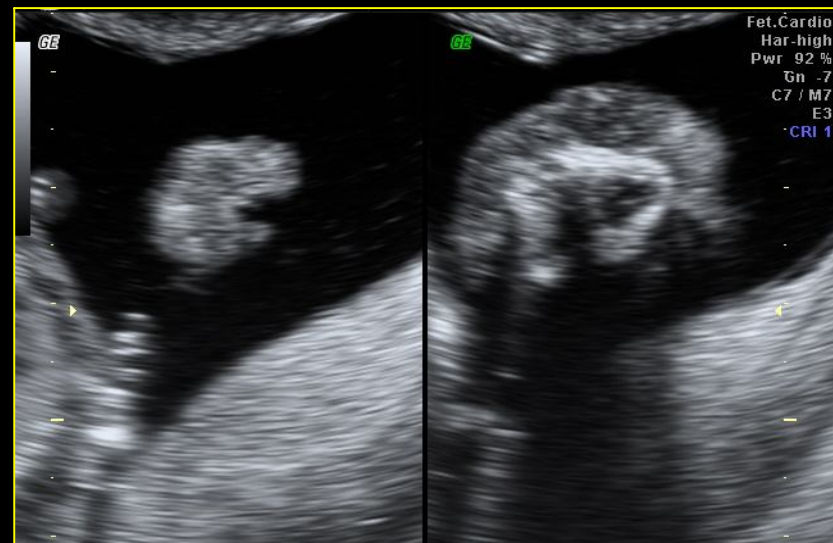




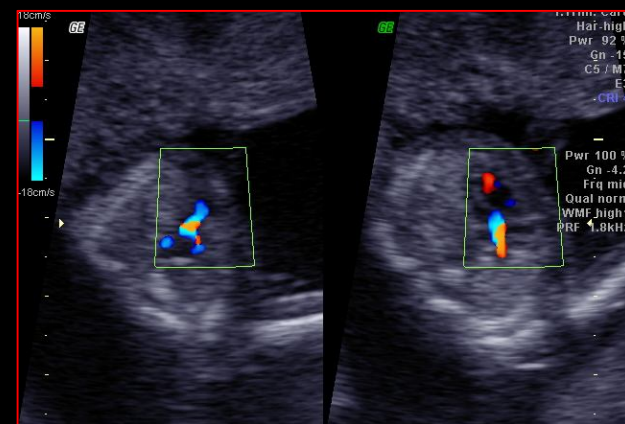
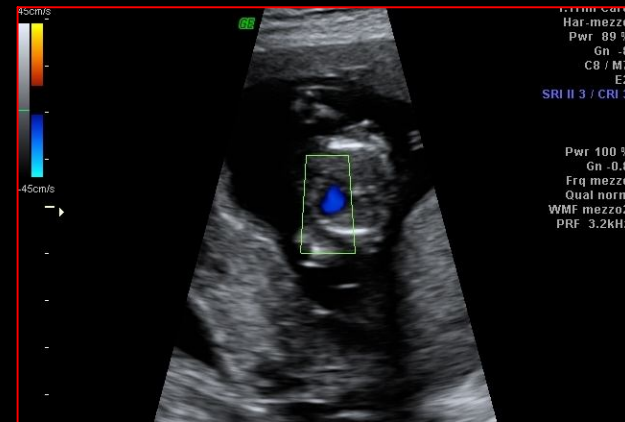
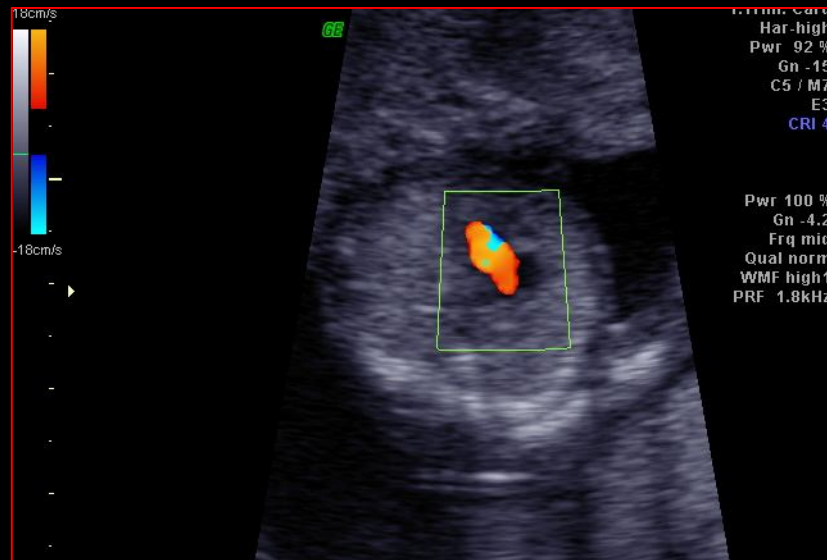
Valutazione Doppler nel I trimestre



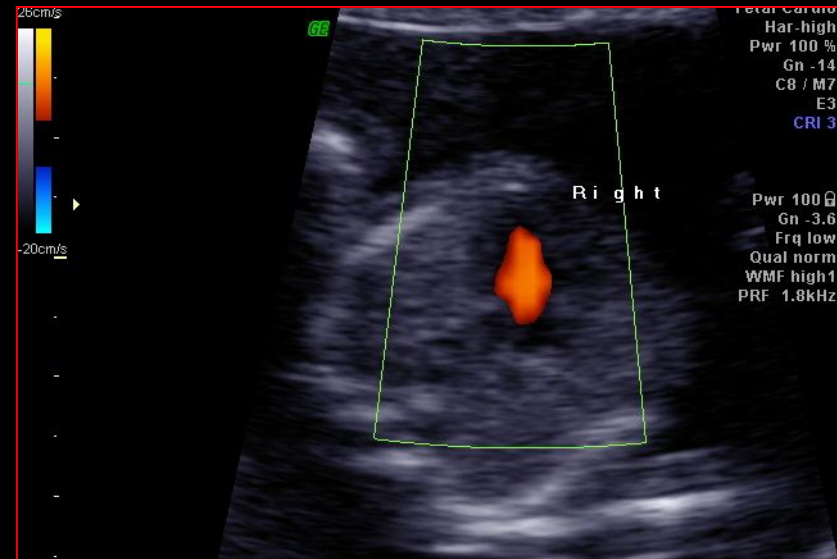
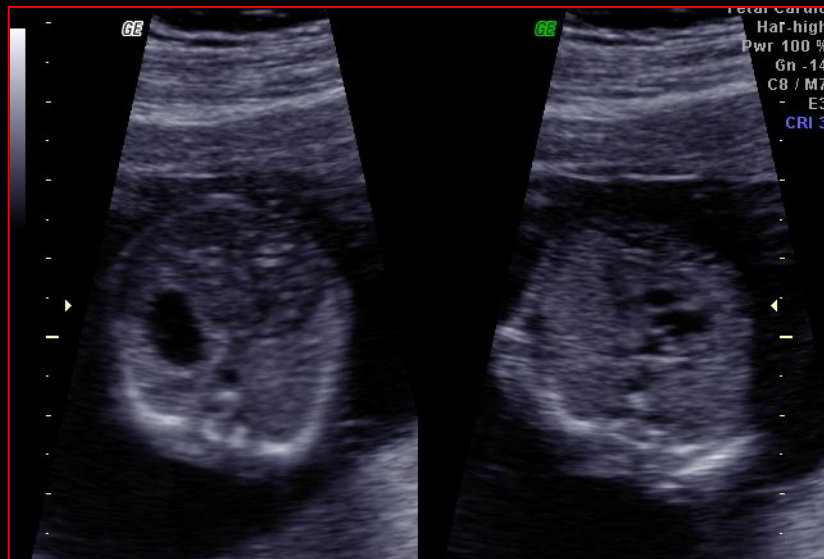
Malformazioni fetali nel I TRIMESTRE



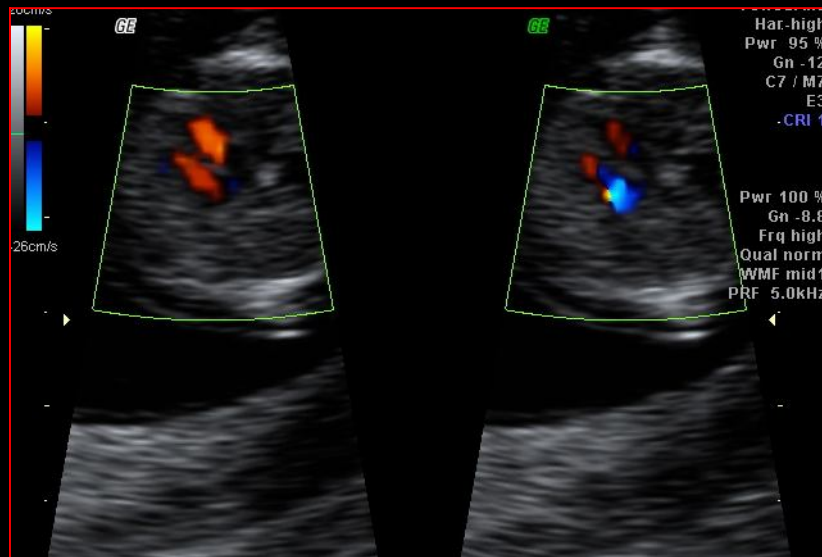
Canale atrioventricolare

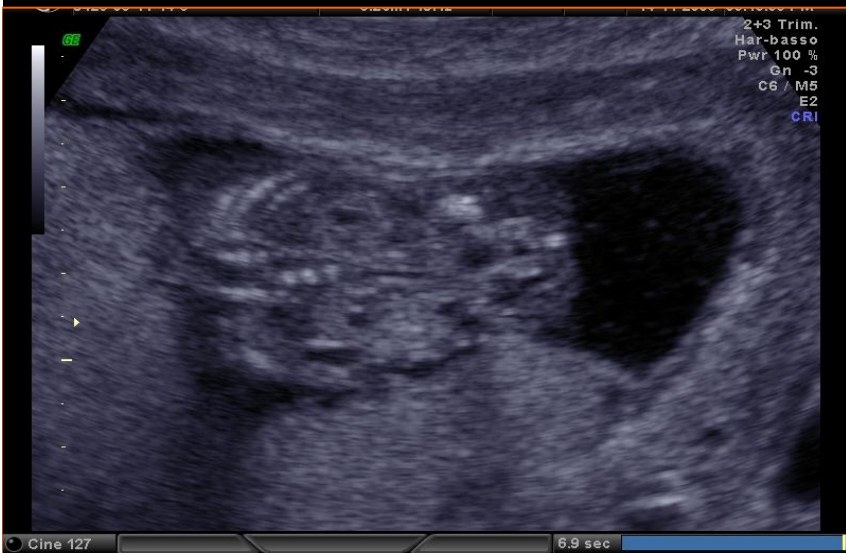


Cuore sinistro ipoplasico



Insufficienza tricuspide





Anomalia della colonna vertebrale

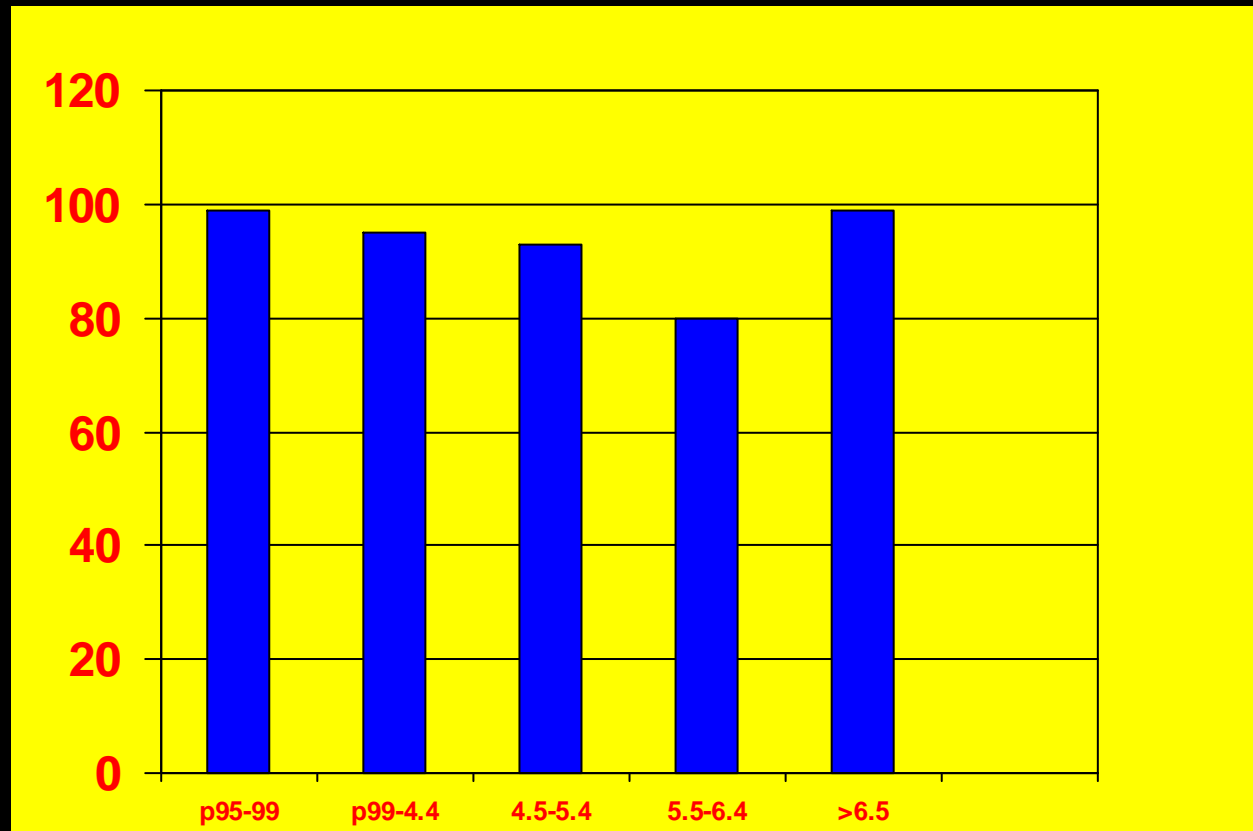


Anomalia degli arti



Outcome favorevole dopo ecografia morfologica normale

Bilardo et al., 2007



CONCLUSIONI

- Le indagini prenatali invasive, le ecografie dettagliate con ecocardiografia, i test genetici e lo screening infettivologico consentono nella maggior parte dei casi di definire l'outcome di una gravidanza
- Se l'indagine ecografica della 20a settimana non ha rilevato anomalie fetali, la probabilità di difetti congeniti alla nascita è uguale alla popolazione generale, indipendentemente dal valore iniziale della NT
- Feto di sesso maschile con $95^{\circ} < NT > 99^{\circ}$ esito migliore rispetto alla femmina

Grazie.....

