Prenatal Testing – Nuchal Translucency and Beyond – What Does it Mean?
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No Relevant Disclosures
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What Does it Mean?

• The idea of nuchal translucency evaluation is part of the greater 11-13 6/7 week ultrasound evaluation of the fetus
• Fetal NT along with maternal age can detect about 75% of trisomic fetuses, if one accepts that 5% of all pregnant women and 100% of high-risk will agree to undergo amniocentesis
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- Adding the pregnancy-associated plasma protein – A (PAPP-A) and free B-human chorionic gonadotropin (hCG) detects 85-90% of trisomic fetuses
• Absent nasal bone is a finding in trisomic fetuses (60-70% of Down syndrome fetuses)
• Adding it as indication for amniocentesis increases diagnosis to 95%
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• Other early screening tests (diagnostic?) included fetal cell evaluation from maternal peripheral blood and more recently cell-free fetal DNA in maternal peripheral blood
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• The late first trimester exam is also critically helpful in determining viability, gestational age, major fetal anomalies and, in twin gestations, chorionicity

Timor-Tritsch. OBG Management; 24,12: 37-45
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• Importantly it will confirm the pregnancy location in most cases
• With the increasing Cesarean rate, there are increasing cases of Cesarean-scar pregnancies and placenta accreta.
• This allows for treatment with chemotherapy, if caught early enough

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• New criteria exist for predicting pregnancy failure based on early ultrasound

• Using an observational cross-sectional study design with prospective data, the following are suggested to diagnose failed pregnancy;
  – Mean sac diameter (MSD) >21 mm without yolk sac or embryo
  – Absent heart beat with CRL ≥ 5.3 mm

• These are slightly more conservative

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- Cranial bones
- Ventriculomegaly
- Holoprosencephaly
- ICT related to spina bifida
- 4-chamber heart and arches
- Stomach
- Bladder
- Kidneys
- Renal arteries
- Umbilical cord insertion is a pitfall
- Extremities – especially long bone number
- Spine

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- ICT, marker for spina bifida, is the subject of a prospective study by Tulin Ozcan that is being coordinated through the North American Fetal Therapy Network (NAFTNet)
- Clearly an expanding horizon
There are new concerns to be aware of in the risk assessment for aneuploidy using 1\textsuperscript{st} trimester NT.

The thought that the second trimester LR for aneuploidy was accurate was based on the independence between 1\textsuperscript{st} and 2\textsuperscript{nd} trimester markers.

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- In fact, a correlation between log NT – MoMs and all 2nd trimester continuous variables as well as nuchal edema and hyperechogenic bowel was found.
- It is already known that cardiac malformations are more common with increasing log NT – MoMs.

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• It is already known that cardiac malformations are more common with increasing NT
  

• Pathology study showed
  - NT ≤ 3 mm 1/11 had AVSD
  - NT ≥ 4 mm 19/25 had AVSD
  

• However, NT is not associated with a higher rate of cardiac defects in known Trisomy 21 fetuses in a prospective study using ultrasound
  
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A comment on noninvasive prenatal testing (NIPT) using cell-free DNA (cf DNA)

• The first study using a routinely screened population was just published
• All previous studies looked at high-risk populations
• 1939 euploid pregnancies had risk scores for trisomies 21 and 18 <0.01% in 99.85% of cases
• 8 cases of trisomy 21 and 2 cases of trisomy 18 had risk score > 99% with < 0.01% for the other

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• When examining some of the studies on predicting Trisomy 21, elective terminations were 90%.
• Same holds true for studies examining nuchal thickness, heart defects (mostly AVSD).
• All prior to validating the effects of the findings on the health of the newborn.
• In populations, geographic or demographic, where termination is not as commonly practiced, cf DNA is an important tool.
The meaning of all these “tests” is enhanced information

• Many moms desire a greater level of information prior to making any decisions regarding termination or treatment and they are savvy to the risks of invasive testing – consumerism in medicine

  ➢ Better knowledge of 3D volume acquisition
  ➢ The certainty of cf DNA
  ➢ The availability of early MRI
  ➢ The growing expertise and accuracy of early fetal echocardiography
  ➢ The availability of Maternal-Fetal Medicine specialists who are willing to take on these patients
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This field is clearly at risk if evaluated from a financial cost-benefit analysis

• Dichotomous views of the fetus – patient or property
• Generates competing or contradictory views
• There are no honest forums for this discussion at the payer level – either public or private

Emotional leverage keeps all testing available at this time
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