Fetal Care Center of Cincinnati

Comprehensive Diagnosis & Treatment

# Prenatal Testing – Nuchal Translucency and Beyond – What Does is Mean?

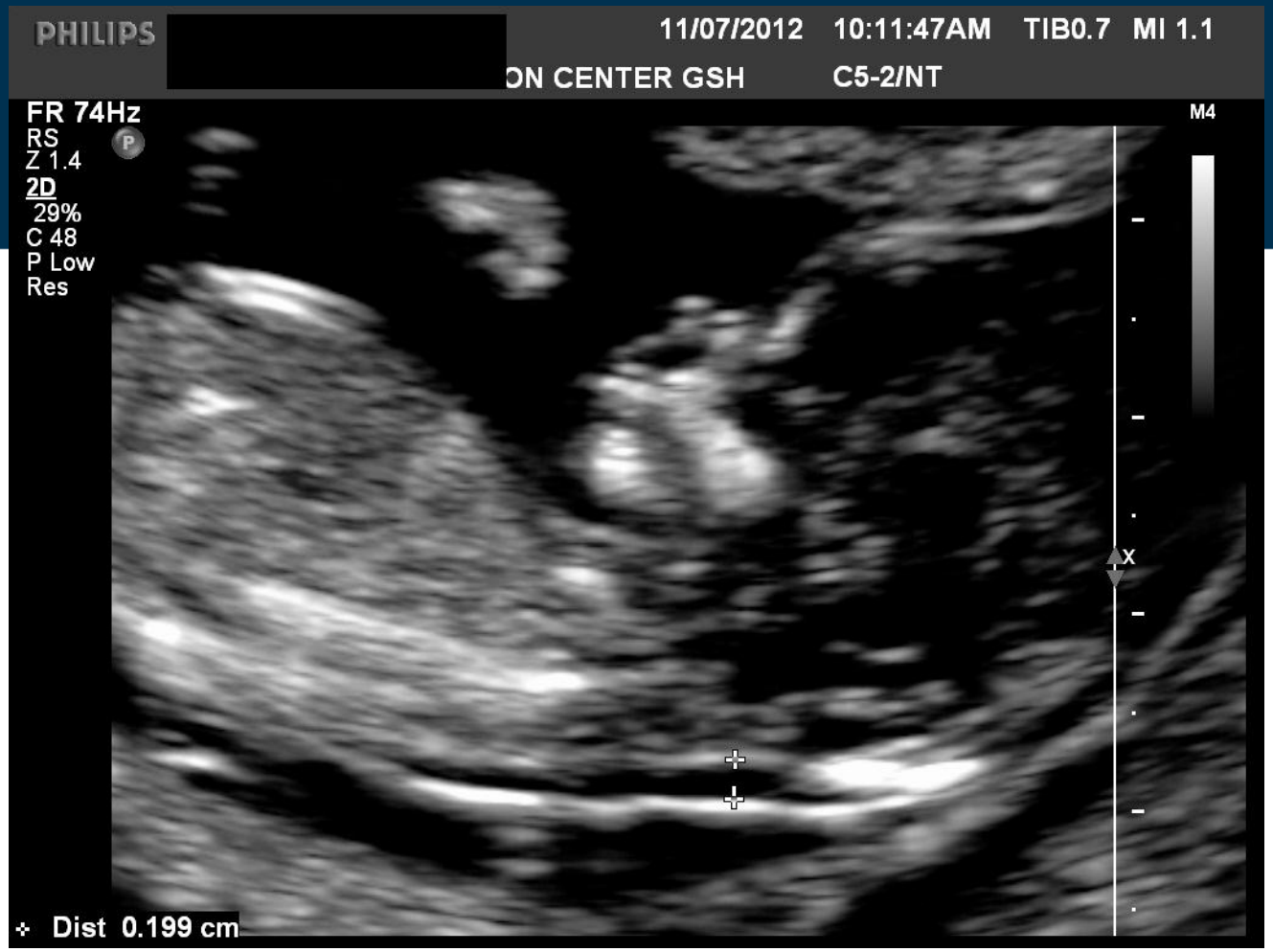
# Prenatal Testing – Nuchal Translucency and Beyond
# What Does is Mean?

William J. Polzin, M.D.
Co-Director, Fetal Care Center of Cincinnati
Director, Division of Maternal-Fetal Medicine
Good Samaritan Hospital
Cincinnati, OH

# No Relevant Disclosures

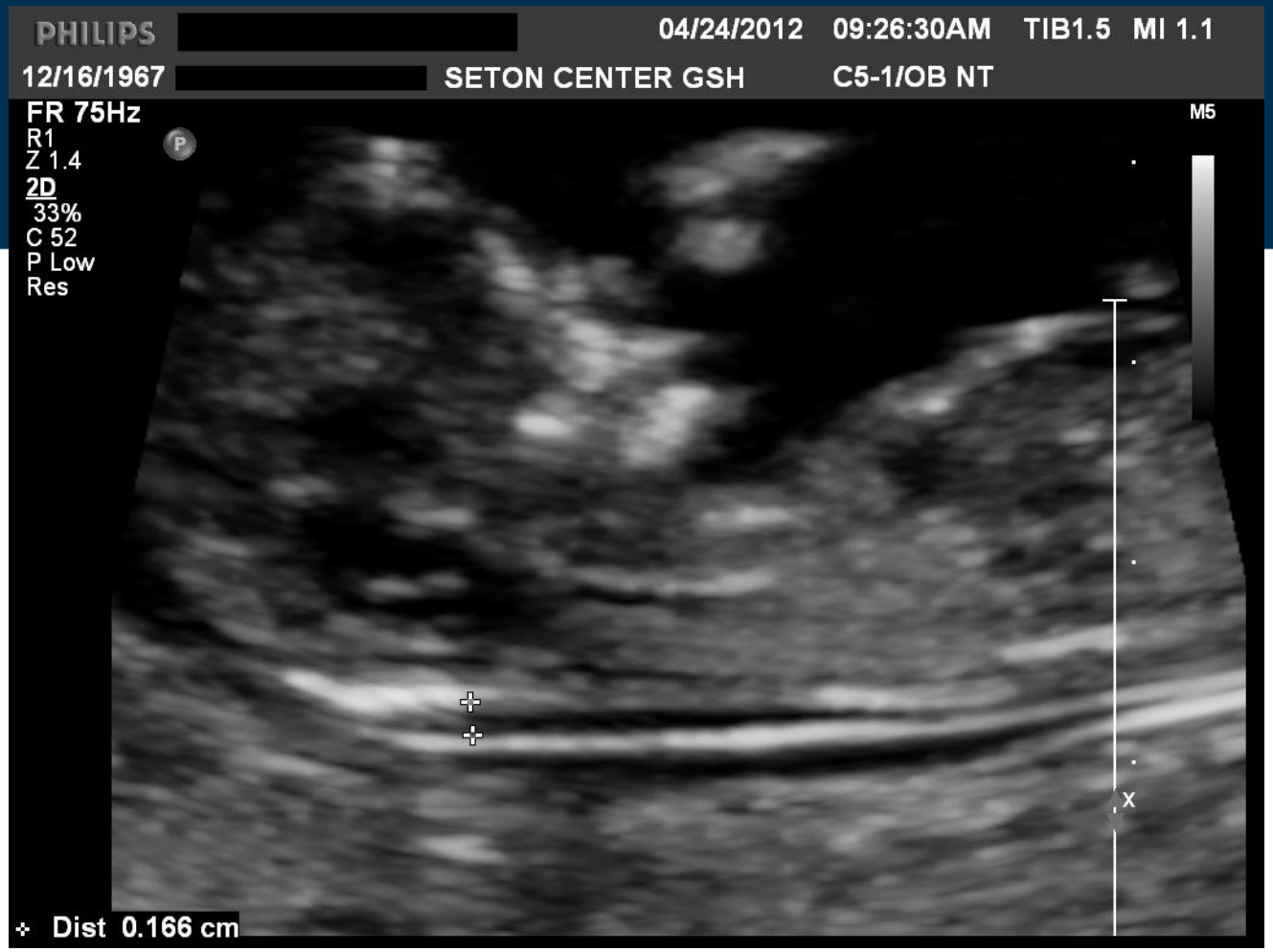
# Prenatal Testing – Nuchal Translucency and Beyond
# What Does is Mean?

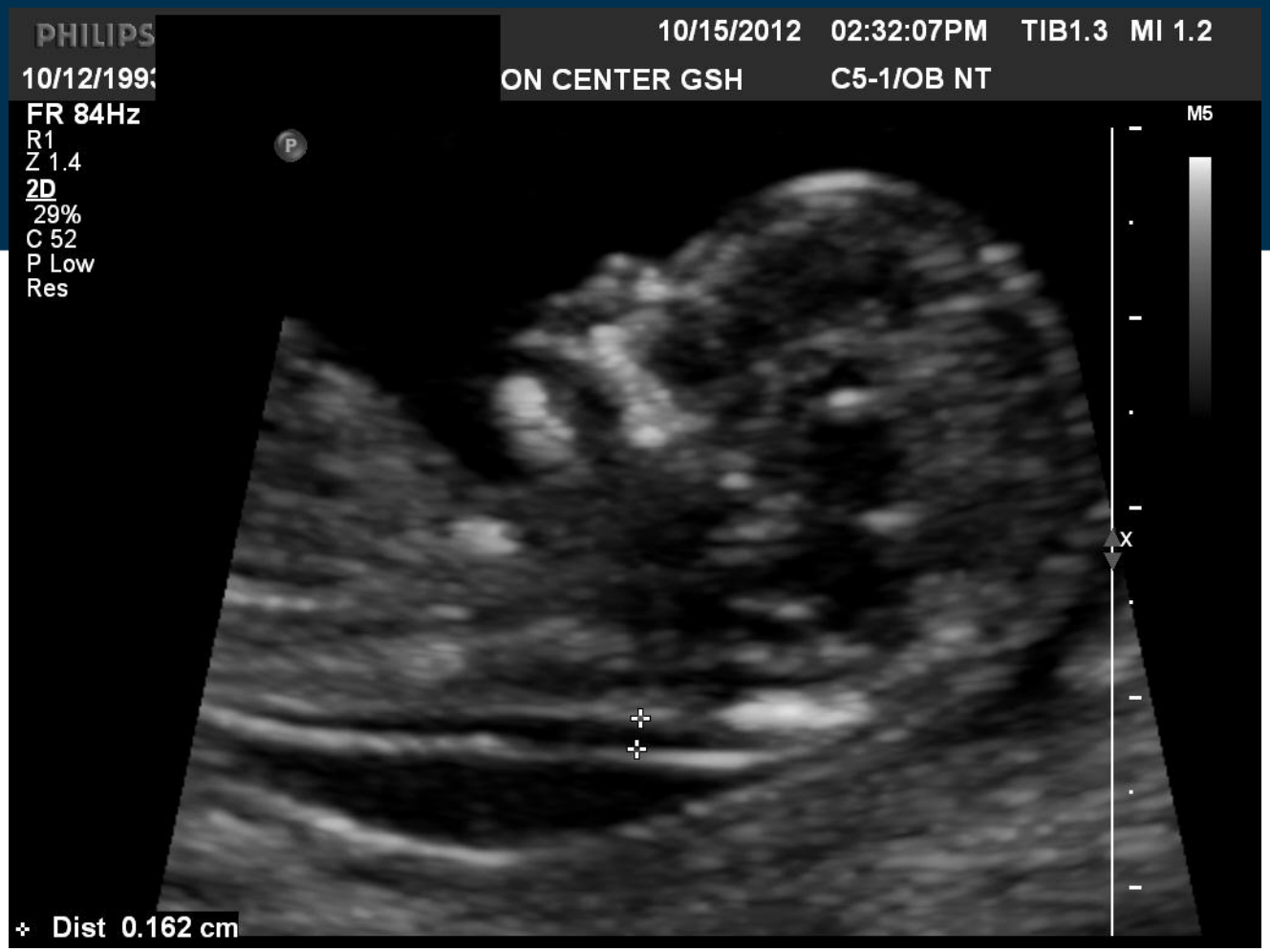
- The idea of nuchal translucency evaluation is part of the greater 11-13 6/7 week ultrasound evaluation of the fetus


PHILIPS
[redacted]
11/07/2012 10:11:47AM TIB0.7 MI 1.1
ON CENTER GSH C5-2/NT
FR 74Hz
RS
Z 1.4
2D
29%
C 48
P Low
Res
M4
X
Dist 0.199 cm

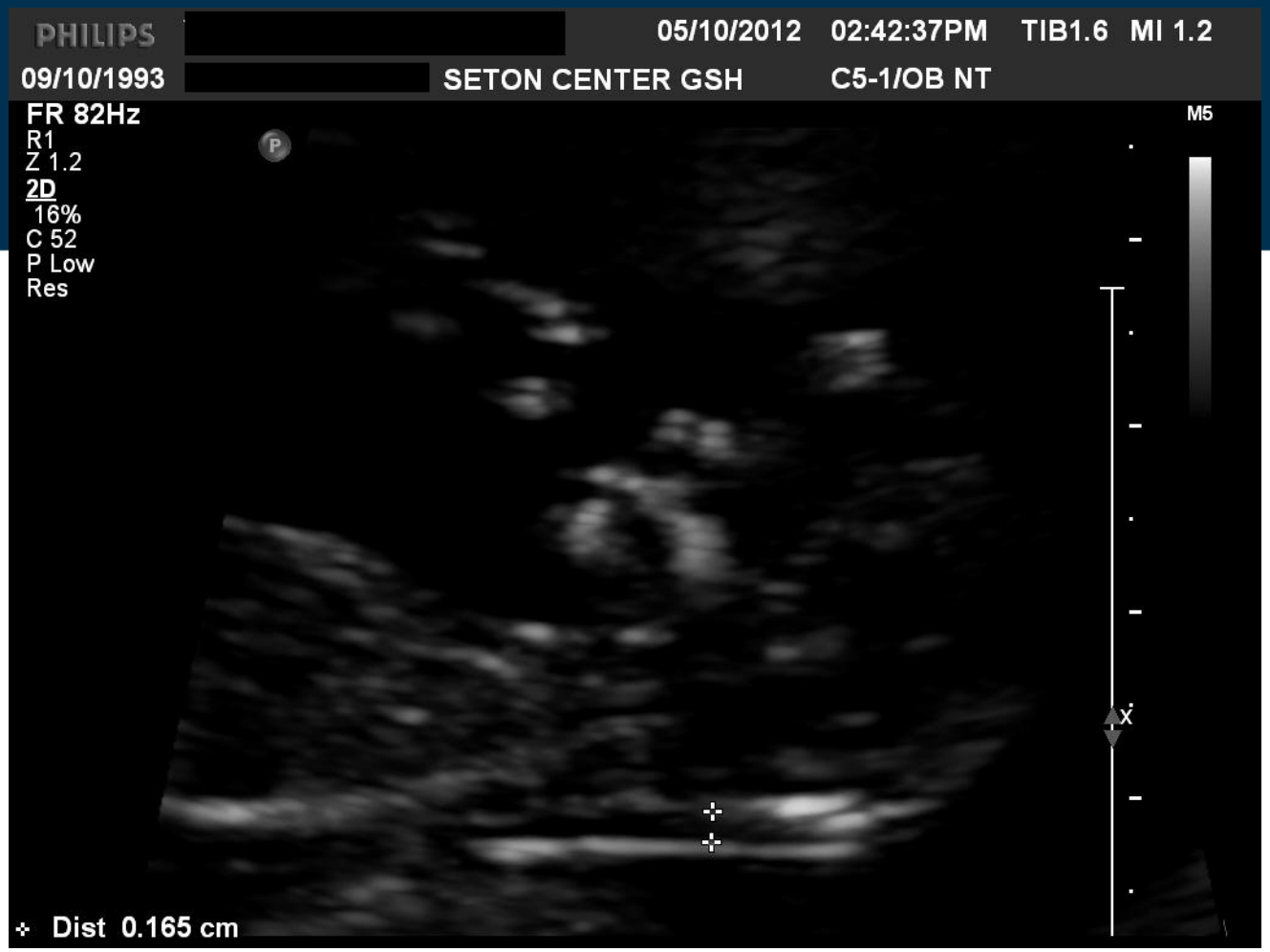
# Prenatal Testing – Nuchal Translucency and Beyond
# What Does is Mean?

- 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


PHILIPS
04/24/2012 09:26:30AM TIB1.5 MI 1.1
12/16/1967
SETON CENTER GSH
C5-1/OB NT
FR 75Hz
R1
Z 1.4
2D
33%
C 52
P Low
Res
M5
Dist 0.166 cm




Fetal Care Center of Cincinnati
CINCINNATI CHILDREN'S • GOOD SAMARITAN • UNIVERSITY HOSPITAL

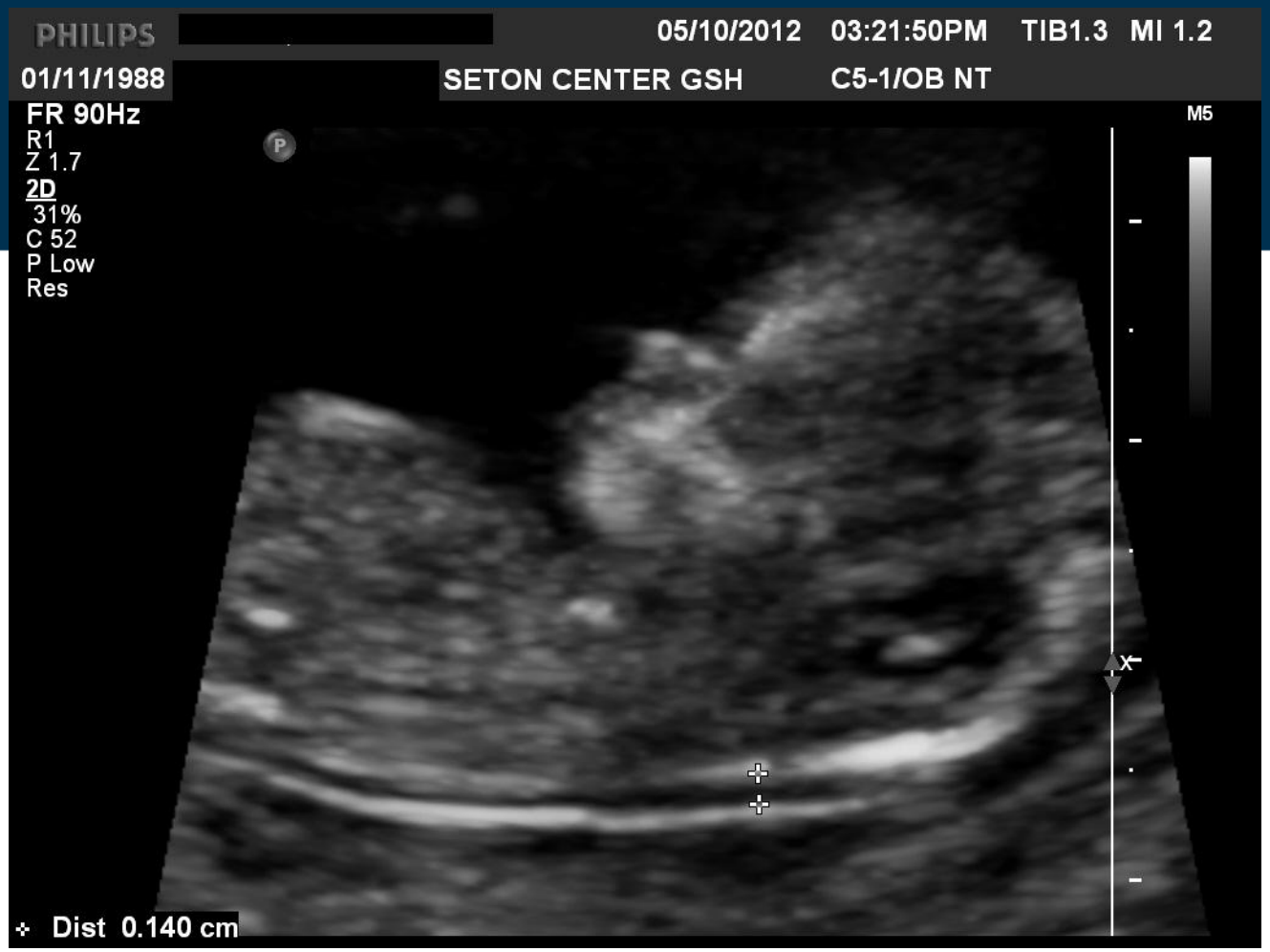

PHILIPS
05/10/2012 02:42:37PM TIB1.6 MI 1.2
09/10/1993
SETON CENTER GSH
C5-1/OB NT
FR 82Hz
R1
Z 1.2
2D
16%
C 52
P Low
Res
M5
Dist 0.165 cm

# Prenatal Testing – Nuchal Translucency and Beyond
# What Does is Mean?

- Adding the pregnancy-associated plasma protein – A (PAPP-A) and free B-human chorionic gonadotropin (hCG) detects 85-90% of trisomic fetuses

# Prenatal Testing – Nuchal Translucency and Beyond
# What Does is Mean?

- 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%


PHILIPS
05/10/2012 03:21:50PM TIB1.3 MI 1.2
01/11/1988
SETON CENTER GSH C5-1/OB NT
FR 90Hz
R1
Z 1.7
2D
31%
C 52
P Low
Res
M5
Dist 0.140 cm

# Prenatal Testing – Nuchal Translucency and Beyond
# What Does is Mean?

- Other early screening tests (diagnostic?) included fetal cell evaluation from maternal peripheral blood and more recently cell-free fetal DNA in maternal peripheral blood

# Prenatal Testing – Nuchal Translucency and Beyond
# What Does is Mean?

- 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

# Prenatal Testing – Nuchal Translucency and Beyond
# What Does is Mean?

- 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

Timor-Tritsch. OBG Management 2012; 24,12: 37-45

# Prenatal Testing – Nuchal Translucency and Beyond
# What Does is Mean?

- 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) $\geq$21 mm without yolk sac or embryo
  - Absent heart beat with CRL $\geq$ 5.3 mm
- These are slightly more conservative

Abdallah. Ultrasound Obstet Gynecol 2011; 38: 497-502

# Prenatal Testing – Nuchal Translucency and Beyond
# What Does is Mean?

- 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

Timor-Tritsch. OBG Management 2012; 24,12: 37-45

# Prenatal Testing – Nuchal Translucency and Beyond
# What Does is Mean?

- 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)
- Other excellent review articles on this topic include Syngelaki. Prenat Diagnosis 2011; 31(1): 90-102 and Fong. Radiographics 2004; 24(1): 157-174
- Clearly an expanding horizon

# Prenatal Testing – Nuchal Translucency and Beyond
# What Does is Mean?

- There are new concerns to be aware of in the risk assessment for aneuploidy using 1st trimester NT.
- The thought that the second trimester LR for aneuploidy was accurate was based on the independence between 1st and 2nd trimester markers

Miguelez. Ultrasound Obstet Gynecol 2012; 39: 274-278

# Prenatal Testing – Nuchal Translucency and Beyond
# What Does is Mean?

- 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

Miguelez. Ultrasound Obstet Gynecol 2012; 39: 274-278

# Prenatal Testing – Nuchal Translucency and Beyond
# What Does is Mean?

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

  Atzei. Ultrasound Obstet Gynecol 2005; 26: 154-157

- Pathology study showed
  - NT $\leq$ 3 mm  1/11 had AVSD
  - NT $\geq$ 4 mm 19/25 had AVSD

  Hyett. Am J Obstet Gynecol 1995: 172: 1411-1413

- However, NT is not associated with a higher rate of cardiac defects in known Trisomy 21 fetuses in a prospective study using ultrasound

  Mogra. Ultrasound Obstet Gynecol 2011; 38: 320-324

# Prenatal Testing – Nuchal Translucency and Beyond
# What Does is Mean?

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

Nicolaides. Am J Obstet Gynecol 2012; 207: 374-376

# Prenatal Testing – Nuchal Translucency and Beyond
# What Does is Mean?

- 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

# Prenatal Testing – Nuchal Translucency and Beyond
# What Does is Mean?

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

# Prenatal Testing – Nuchal Translucency and Beyond
# What Does is Mean?

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

# Prenatal Testing – Nuchal Translucency and Beyond
# What Does is Mean?

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