

Prenatal Screening Items to Consider in Discussion with Patient

- Prenatal screening is not genetic screening. It is screening for 3 conditions (Down syndrome, Trisomy 18, and open neural tube defects) that can occur to any woman in any pregnancy, and it has been recommended nationally that all women be offered screening for these conditions. The patient has a choice whether to have screening or decline.
- If there is a specific risk factor for the woman (abnormal ultrasound, family history of birth defects or genetic conditions, multiple miscarriages), offer genetic counseling and/or diagnostic testing. For all others offer some form of screening.
- Screening tests estimate the chance a baby might have one of these conditions. They cannot say for certain. No test can say a baby is normal.
- Increased NT can be seen with chromosome abnormalities and some heart defects. Thus, NT measurement can also indicate possible congenital heart defect.
- We recommend Integrated Screening as it has the highest detection rate.

Approximate Detection Rate in %

Screening Method	DS	T18	ONTD
FTO	85	80	0
IS	87	90	80
SIS	86	90	80
Quad	80	80	80

- The reason to have a screening test is if the patient feels the information might be helpful to her in some way. Further diagnostic tests that find an abnormality will usually trigger a decision to terminate the pregnancy or continue the pregnancy with no specific treatment. However finding ONTD, T18 or a congenital heart defect can be helpful in pregnancy management if the decision is to continue the pregnancy.

Definitions:

FTO – First Trimester Only: NT @ 12-13 weeks (all gestational ages approximate), Blood at 12-13 weeks. Patient will need to be offered AFP at 16 weeks.

IS - Integrated Screening: NT at 12 weeks, Blood at 12 weeks, Blood at 16 weeks.

SIS - Serum Integrated Screening: Blood at 12 weeks, Blood at 16 weeks (can be helpful when NT is not available).

Quad -Quad screening: Blood at 16 weeks.

NT - Nuchal Translucency: Ultrasound measurement of space behind baby’s neck. To use the measurement in IS, it must be performed by a certified ultrasonographer.

DS – Down syndrome.

T18 – Trisomy 18.

ONTD – Open neural tube defect.