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Options for Prenatal Diagnosis – Just the Basics

Genetic Counseling

Timing- variable, depending on the indication.

Mandatory prior to any invasive testing. Consultation is non-directive and includes risk assessment (including complete family and pregnancy history), education, and help in choosing prenatal tests appropriate for the patient. Studies show up to 50% higher detection for hereditary disorders when history is taken by genetic counselor rather than by an OB.

Nuchal Translucency (NT)

Timing - optimally **11 to 13w6d**.

Performance and interpretation of the test requires certification by Fetal Medicine Foundation. Calculates a statistical risk for Down syndrome based upon maternal age, GA, history of previous child with DS, and NT. Useful for trisomy 13, 18 and Turner's syndrome. Sensitivity ranges from 72-80% among certified centers, with a 5% positive test rate.

Combined screen (NT plus first trimester serology)

Timing – The serology can be done 10w0d to 12w6d and the NT timing is as above. The blood test (only at Dynacare) can be done before, during or after the NT scan. Sensitivity estimated at 85-91% with a 3-5% positive test rate. Useful for trisomy 18.

Performance issues: Labwork performed at Dynacare - can be completed at time of NT or by referring OB as early as 10 weeks with rest of paperwork to be completed at time of NT. Results of serology and NT will be combined with maternal age to give an adjusted risk for Down syndrome approximately one week after the blood is drawn. Risk estimates for neural tube defects (i.e. spina bifida) are not given by the combined screen, thus maternal serum AFP needs to be drawn at 15-22 weeks. Not useful for Down syndrome risk estimate with multiples, does not screen for trisomy 13, 47XXX or 47XXY

Integrated Screen (NT integrated with both first and second trimester serum biochemistry.)

Timing – Serum PAPP-A (IPRP1) at **10w0d-12w6d** (based upon CRL measurements), **NT at 11w0d-13w6d**, and **Quad Screen (IPRP2) 15 to 20 weeks**. Sensitivity estimated at 80-85% for Down syndrome with a 1% positive test rate.

Performance issues: Labwork performed at Dynacare - IPRP1 to be sent first – this can be completed at time of NT or by referring OB as early as 10 weeks with rest of paperwork to be completed at time of NT. Paperwork given to patient to have IPRP2 drawn with instructions about timing. Results of IPRP1 and IPRP2 and NT will be integrated with maternal age to give an adjusted risk for Down syndrome. Maternal serum AFP is used to estimate risk of open neural tube defects. MS-AFP, estriol, and HCG are used to estimate risk of trisomy 18. Not useful for Down syndrome risk estimate with multiples, does not screen for trisomy 13, 47XXX or 47XXY

Chorionic Villus Sampling (CVS)

Timing – **10 to 14 weeks**

Performance issues – 0.5-1% incidence of mosaicism, or growth failure. 95% of procedures performed transabdominally.

Definitive test, useful for first trimester diagnosis of aneuploidy; does not test for neural tube defect so serum AFP is recommended at 17 weeks and targeted high-resolution ultrasound at 18 weeks. Excessive risk of miscarriage 1%; no increased risk of limb defects if performed after 10w0d

Quad Screen

Timing – serum test at **15 to 20 weeks**

Maternal serum levels of AFP, hCG, UE3, and Inhibin A as well as maternal age, weight, and family history give a readjusted risk for Down syndrome, trisomy 18, and neural tube defects; sensitivity of 70% for Down syndrome with a 5% test positive rate.

Performance issues: Not useful for risk estimates with multiples, does not screen for trisomy 13, 47,XXX or 47,XXY

Targeted High Resolution Ultrasound

Timing – usually **16 to 22 weeks**, or at least one week *after* the Quad or Integrated Screen test (IPRP2) is drawn.

Diagnostic test to assess any structural abnormalities or “soft signs” for Down syndrome including nuchal thickening, echogenic bowel, short limbs, pyelectasis, with a 50% - 60% sensitivity for detection of Down syndrome. Baseline age, quad or integrated screen adjusted risk can be adjusted up or down depending upon the findings .

Amniocentesis

Timing – optimally **16 to 18 weeks** though can be performed at 15 to 20 weeks

Detects 99.5% of all chromosome abnormalities; full karyotype results usually within two weeks. In cases of high risk from serum screening, frank anomalies by ultrasound, or advanced gestation we can also order Fluorescence In Situ Hybridization (FISH), which will detect aneuploidy of chromosomes 13, 18, 21, X, and Y with results in 24-48 hours. Assesses AFAFP levels to screen for neural tube defect and abdominal wall defects. Definitive invasive diagnostic test with up to 1% excessive risk of miscarriage

Fetal Echocardiogram

Timing – usually **20 – 22 wks**

Diagnostic test to assess detailed cardiac anatomy in patients at high risk for cardiac anomalies eg: from IDDM, medication exposure, structural anomaly, or arrhythmia. Performed by Pediatric Cardiologist or Perinatologist. Sensitivity quite variable depending upon lesion and examiner.