Risk Assessment for Birth Defects

While fortunately most babies are born healthy, approximately 3-5% of babies are born with a major birth defect such as a heart problem, chromosomal disorder, or a neural tube defect such as spina bifida. Some of these are more common in older mothers, but many are not age-related. Many of these are detectible during pregnancy and knowledge of these problems significantly impacts appropriate prenatal care and delivery. Several tests are available for determining the risk of your baby having a serious problem.

FIRST TRIMESTER SCREENING

**Ultrascreen:** First trimester ultrasound including nuchal translucency (an ultrasound measurement of the fluid at the back of the baby’s neck), nasal bone, etc. with a blood test for PAPP-A and HCG (some centers also include an early AFP). This will detect approximately 90% of babies with Down syndrome and 98% of babies with Trisomy 18. It also screens for elevations in risk for pre-eclampsia, growth restriction, heart defects, etc. This is performed between 11-13 weeks of pregnancy.

**Maternal Serum AFP:** Blood test performed anytime between 15-20 weeks to determine the risk of a neural tube defect, ventral wall defects, etc.

**Detailed anatomy ultrasound:** Performed between 18-20 weeks to assess the baby’s anatomy. Usually the sex of the baby can be seen on this scan, if desired.

SECOND TRIMESTER SCREENING with BLOODWORK

**QUAD screen/Maternal serum screen:** A blood test performed at 15-20 weeks to determine the risk of some chromosomal abnormalities such as Down syndrome or Trisomy 18 or 13 or neural tube defects. This blood test has a lower detection rate for chromosomal abnormalities than the ultra-screen.

**Detailed anatomy ultrasound:** Performed between 18-20 weeks to assess the baby’s anatomy. Usually the sex of the baby can be seen on this scan, if desired.

ADDITIONAL TESTING OPTIONS

**NIPT:** non-invasive prenatal testing which is available for high risk screened individuals. The blood test picks up circulating cell, free fetal and DNA in the maternal blood.

**Chorionic Villus Sampling (CVS):** CVS involves inserting a tiny tube into the uterus and taking a piece of the placenta. The placental tissue is cultured and the baby’s chromosomes are determined. This test does not check for neural tube defects. It is typically done at 12-13 weeks of pregnancy.

**Amniocentesis:** Amniocentesis involves inserting a needle into the uterus and withdrawing a small amount of fluid from around the baby. Both chromosomal determination and testing for neural tube defects can be obtained from the fluid. It is typically done at 16-20 weeks of pregnancy.
Benefits of determining the presence of a major birth defect:

If your baby is found to have a major birth defect, knowing about it during the pregnancy can help you and your family to plan and make decisions about your options. For instance, infants with Down syndrome are at increased risk for major defects of several organ systems. Women with babies known to have a major birth defect such as a heart problem, gastrointestinal abnormality or spina bifida, for example, may not be able to deliver at Howard County General Hospital, but may need to deliver at a tertiary care center such as University of Maryland or Johns Hopkins Hospital where specialists are immediately available. Often, these babies are critically ill or need surgery right after delivery and it is much better for the baby to be delivered at the hospital where they will receive the appropriate care, rather than be separated from their mother and transported there after delivery while critically ill.

I have read the above and request:

_______ 1st trimester screening with NT scan, AFP, & detailed anatomy scan

_______ 2nd trimester screening with quad screen and anatomy scan

__________________________________________  ______________________________
Patient signature and date  Witness

PLEASE VERIFY INSURANCE COVERAGE
FOR ANY PROCEDURES BEFORE SCHEDULING