Chromosome Testing at Emerson Hospital Maternal Fetal Medicine

What is chromosome screening?

- Everyone is offered the chance to determine whether their pregnancy has an atypical chromosome makeup
- Chromosomes are the large structures that contain DNA. Pieces of DNA within chromosomes are called genes. A human cell typically has 46 chromosomes, in 23 pairs
- Detecting pregnancies with chromosome conditions allows families to be informed and plan their care. Not everyone chooses to test for chromosome conditions during pregnancy
- An example of a condition caused by a difference in chromosome number is Down syndrome, which is due to an extra copy of chromosome 21. Some chromosome conditions can impact children's health and learning
- Typically chromosome conditions occur by chance and are not inherited. There is typically *not* a family history of a chromosome condition

What chromosome screening tests are available?

Chromosome screening tests give an estimate of risk for a chromosome condition, but not a diagnosis.

- **Ultrasound** – uses images of the pregnancy to follow baby's growth and development. Not all chromosome conditions will have findings on ultrasound
- **Cell-free DNA (cfDNA)** – detects pieces of the genetic code, called DNA, that come from the placenta and are in the patient's blood. This testing is performed after 10 weeks of pregnancy

What diagnostic procedures are available?

- **Chorionic villus sampling (CVS)** – a small sample of the placenta is removed and sent for testing. This procedure is performed at Brigham and Women's Hospital
- **Amniocentesis** – a small sample of the amniotic fluid around the fetus is removed and sent for testing.
- Both CVS and amniocentesis obtain samples directly from the pregnancy using a needle under ultrasound-guidance. These tests have very small risks of pregnancy complications.

What conditions are included in cfDNA screening?

- trisomy 21 (Down syndrome)
- trisomy 13 and trisomy 18
- optional sex chromosome analysis which can identify conditions such as Turner syndrome and Klinefelter syndrome

What can diagnostic tests detect?

- A **karyotype** can detect an extra or missing copy or large piece of any chromosome including chromosomes 13, 18, 21, X and Y
- A **microarray** can detect extra or missing pieces of chromosomes too small to find with cfDNA or karyotype

Diagnostic tests can thus detect trisomies and also syndromes such as DiGeorge syndrome, Cri-du-Chat syndrome, Williams syndrome and some associated with autism or intellectual disability

What about fetal sex information?

Sex chromosome screening and testing are available but optional. Sex chromosomes do not always correlate with gender

- cfDNA screens for conditions with atypical numbers of the sex chromosomes (X and Y) but with less accuracy than it screens for other chromosomes (13, 18, 21). It predicts the fetal sex with high accuracy
- Diagnostic testing can include analysis of the sex chromosomes with definitive results

What test is right for me?

You can discuss the options with your provider or a genetic counselor. Consider when deciding what's right for you:

- Would you want to know if the pregnancy has a chromosome condition?
- Would you prefer a screening test (no risk to the pregnancy and results reduce risks for some common chromosome conditions) or a diagnostic test (small procedure-related risks and results provide a definitive answer about many chromosome conditions)

How much do these tests cost?

- Insurance companies typically cover the cost of cfDNA, CVS or amniocentesis.
- The genetic counselor can provide additional information about estimates for cost or insurance coverage. When cfDNA is not covered by insurance, the cost is $349. Patients with Masshealth do not receive a bill for cfDNA.