Genetic Testing in the first trimester

Chorionic Villus Sampling (CVS) placental biopsy

For karyotype and/or direct DNA and cytogentetic testing (chromosomal)

- Test is done between 10 & 14 weeks gestation
- Conducted through the cervix or through the abdomen
- A sample is taken from the placental tissue for testing
- Invasive procedure with .2% increased risk for miscarriage
- Tests for chromosomal abnormalities, and some genetic abnormalities
- Does not detect neural tube defects
- Only recommended for those that are considered high risk*

Ultrasound

Screen for increased risk of aneuploidy by measuring nuchal translucency

- The test is done between 10 & 14 weeks gestation
- Non-invasive- done with an ultrasound
- The fetus' nuchal fold (back of neck) is measured.
- A measurement of over 3mm can be associated with an increased risk of Trisomy 21 and other chromosomal abnormalities, as well as heart defects and other genetic syndromes.
- The absence of fetal nasal bone is also associated with Trisomy 21 and can be detected via ultrasound
- Recommended for anyone considering an ultrasound, or that is high risk*

Fetal cell-free DNA testing- NIPT (non-invasive prenatal testing)

Screen for trisomy abnormalities and sex chromosome aneuploidy

- The test is done any time after 10 weeks gestation
- Non-invasive. Blood is drawn from the pregnant person and sent to a lab
- The test screens for possible extra or missing copies of the X chromosome and Y chromosome and trisomy disorders. Also identifies the sex of the fetus
- Results have about a 5% false-positive rate
- Recommended for pregnant people that considered high risk*

Genetic Testing in the first trimester

Maternal Serum Screen

Done in conjunction with the Ultrasound to screen for increased risk of of a chromosomal condition such as Trisomy 21, 18 & 13

- The test is done between 9 & 13 weeks gestation
- Blood is drawn from the pregnant person
- Pregnant persons weight & age as well as relevant medical information is used as well

First-trimester testing/screening general information

- High-risk persons are those over 35 years of age with a family history of chromosomal or genetic abnormalities
- Screenings are non-invasive ways to help you decide if further testing that may be more invasive and carry higher risk should be considered
- All screenings and tests are optional. You have every right to refuse any and all screenings/tests
- All screenings and tests come with a risk of false results. When considering testing/screening consider what you will do with the information, and the stress impact the results may have on you and your pregnancy
- Testing/screening can be expensive, not all insurances cover their cost
- Aneuploidy- incorrect number of chromosomes on a cell. Some of the most common are: Trisomy-21 (Down's syndrome), Trisomy-18 (Patau syndrome), Trisomy-18 (Edwards Syndrome)

Resources for further information:

- St Lukes Maternal-Fetal Medicine: (208) 381-3088
- March of Dimes
- Genetic Alliance
- Mayo Clinic- CVS testing

Frye, A. (2007). Understanding Diagnostic Tests in the Childbearing Year: a holistic approach. Labrys Press.
King, T. L., & King, T. L. (2019). Varney's midwifery. Jones & Bartlett Learning.