Amniocentesis

What is an amniocentesis?
Amniocentesis is a procedure performed typically after the 16th week of pregnancy. Amniotic fluid is obtained with a long, thin needle entering through the abdomen into the amniotic sac. Ultrasound guidance is used throughout the procedure to get a pocket fluid away from baby. The fluid is sent to a lab to be tested for chromosomal conditions or single gene conditions like sickle cell disease or Noonan syndrome.

Are there risks associated with amniocentesis?
There is risk for miscarriage, bleeding, infection, etc. It is important to ask the physician performing this procedure what he or she personally quotes.

When would someone consider amniocentesis?
- If someone receives a positive blood test for chromosomal condition
- If someone was considered advanced maternal age
- When a couple are known carriers of the same condition or a genetic female/woman is a carrier of an x-linked condition
- When a birth difference is identified on ultrasound
- If someone transferred an embryo that had a mosaic PGT-A result

*This is not a comprehensive list of reasons for why someone may choose to proceed with an amniocentesis.

What are the limitations of amniocentesis?
Amniocentesis is performed in the second trimester, so diagnostic results are not available until the second trimester which may limit reproductive options. The cells that are assessed come from the baby's skin and digestive system tissues, so it is unable to provide results on every tissue type - which is not always feasible after birth either.

What are the benefits of amniocentesis?
The procedure can help to exclude or confirm a possible diagnosis. If a diagnosis is confirmed, then someone may have the option to make reproductive choices such as adoption, termination, or learning more about the diagnosis to prepare for their baby's arrival. It also is able to test the baby's DNA directly, so the risk of placental mosaicism, where the placental DNA differs from the baby's, is not a typically a concern.

For more information, speak to a healthcare provider.