General information about positive NIPT results

My patient's NIPT is positive for monosomy X (Turner syndrome). What does this mean? Your patient's NIPT result suggests the presence of one X sex chromosome and the absence of a second sex chromosome. NIPT is a screening test; false positives can occur. The actual chance for the pregnancy to have monosomy X depends on many factors, including the patient's clinical and family history.

Next steps to consider: You should discuss the results and the potential clinical implications with your patient. The American College of Obstetricians and Gynecologists and the Society for Maternal-Fetal Medicine state, "All women with a positive cell-free DNA test result should have further detailed counseling and testing and should have a diagnostic procedure before any irreversible action is taken."[1] Confirmation prior to birth can also help with pregnancy and neonatal management.

Please see below for more information about monosomy X and additional resources.

What is monosomy X? Monosomy X is a condition that is caused by having one X sex chromosome and an absent second sex chromosome (one sex chromosome instead of two sex chromosomes).

What are the features of monosomy X? Many pregnancies with monosomy X will result in a pregnancy loss; however, monosomy X is compatible with continued survival and live birth. Females with monosomy X have variable phenotypes. Typically, females with monosomy X have normal intelligence; however, learning disabilities are possible and variable. Some of the common features of monosomy X include heart defects, kidney abnormalities, short stature, congenital lymphedema, and primary amenorrhea.

What is the prevalence of these conditions? Approximately 1 in 2500 females are born with monosomy X. This condition usually happens by chance and is not typically associated with advanced parental age.

What testing could be considered?

- Specialized genetic tests such as karyotyping, fluorescence in situ hybridization (FISH), qPCR and microarray are available to confirm the presence of monosomy X.
- These confirmatory tests are generally performed on cells from chorionic villus sampling (CVS) or amniocentesis during pregnancy, on cord blood or peripheral blood sample after the baby is born, or on products of conception (POC) in case of a miscarriage.
- Ultrasound evaluation may be useful in aiding with a prenatal diagnosis of monosomy X, but a normal ultrasound cannot exclude this condition.

Resources for monosomy X

Turner Syndrome Society of the United States
http://www.turnersyndrome.org/

Turner Syndrome Resource List
http://www.kumc.edu/gec/support/chromoso.html#xo

The information provided in this sheet is based on literature search performed on 11/28/16. This Information Sheet is intended to provide some general overview of the key issues relating to its subject matter. This sheet is not intended to be an exhaustive discussion of the subject covered by the sheet nor should it be used to substitute for the exercise of a Clinical Laboratory or a Healthcare Provider's legal or professional duties relative to interpreting the test results to which this Information Sheet relates. This sheet is also not intended to serve as a recommendation of management. This sheet is not intended to be a substitute for genetic counseling.

Reference:

1. American College of Obstetricians and Gynecologists. Screening for fetal aneuploidy. Practice Bulletin No. 163. *Obstet Gynecol.* 2016;127:e123-e137.

Additional Sources:

Gardner RJM, Sutherland GR, Schaffer LG. *Chromosome Abnormalities and Genetic Counseling.* 4th ed. New York, NY: Oxford University Press; 2012.

Jones KL. *Smith's Recognizable Patterns of Human Malformation.* 5th ed. Philadelphia, PA: W.B. Saunders Company; 1997.