General information about positive NIPT results

My patient's NIPT is positive for XXX syndrome (triple X syndrome). What does this mean? Your patient's NIPT result suggests the presence of an extra copy of the X chromosome. NIPT is a screening test; false positives can occur. The actual chance for the pregnancy to have XXX syndrome 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 XXX syndrome and additional resources.

What is XXX syndrome? XXX syndrome is a condition that is caused by a female having an extra copy of the X sex chromosome (three copies of the X chromosome instead of the usual two copies).

What are the features of XXX syndrome? XXX syndrome is likely to result in livebirth. XXX syndrome is usually not associated with intellectual disability or severe birth defects. Some of the common features of XXX syndrome include delayed speech and motor development. Females with XXX syndrome can be taller than average height. Pubertal development and fertility is usually normal.

What is the prevalence of this condition? Approximately 1 in 1000 females are born with XXX syndrome. This condition usually happens by chance and can be associated with advanced maternal 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 XXX syndrome.
- 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 prenatal diagnosis, but a normal ultrasound cannot exclude this condition. Ultrasound is usually normal with XXX.

Resources for XXX Syndrome

Genetics Home Reference
http://ghr.nlm.nih.gov/condition/triple-x-syndrome

National Organization for Rare Disorders
https://rarediseases.org/rare-diseases/trisomy-x/

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.