General information about positive NIPT results

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

What is XYY syndrome? XYY syndrome is a condition that is caused by a male having an extra copy of the Y sex chromosome (one copy of the X chromosome and two copies of the Y chromosome rather than the usual one copy of each).

What are the features of XYY syndrome? XYY syndrome is likely to result in live birth. Males with XYY syndrome have variable phenotypes. Some of the common features of XYY syndrome include delayed speech and language development and taller stature. There is a slightly increased risk for males with XYY syndrome to have an autism spectrum disorder or learning disability.

What is the prevalence of this condition? Approximately 1 in 1000 males are born with XYY syndrome. This condition usually happens by chance and is not 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 XYY 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 is not expected to be useful in aiding with a prenatal diagnosis of XYY syndrome, as ultrasound is usually normal. A normal ultrasound cannot exclude this condition.

Resources for XYY syndrome

Genetics Home Reference
http://ghr.nlm.nih.gov/condition/47xyy-syndrome

National Organization for Rare Disorders
http://rarediseases.org/rare-diseases/xyy-syndrome/

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.