General information about positive NIPT results

My patient's NIPT is positive for 21 (Down syndrome). What does this mean? Your patient's NIPT result suggests the presence of an extra copy of chromosome 21. NIPT is a screening test; false positives can occur. The actual chance for the pregnancy to have trisomy 21 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 trisomy 21 and additional resources.

What is trisomy 21? Trisomy 21 is a condition that is caused by an extra chromosome number 21 (three copies instead of two).

What are the features of trisomy 21? Although some pregnancies with trisomy 21 end in pregnancy loss, trisomy 21 often can lead to live birth. Individuals with trisomy 21 have variable physical features and intellectual disability. Some of the common features of trisomy 21 include heart defects, low muscle tone, and differences in facial features. In addition, cognitive impairment can range from mild to severe. People with Down syndrome have a higher risk for certain medical conditions, such as hearing problems, thyroid problems, childhood leukemia, and Alzheimer's disease. People with Down syndrome may require supervision throughout their lives. However, many people with Down syndrome are increasingly attending school and holding jobs. Many individuals with Down syndrome live into adulthood.

What is the prevalence of this condition? Trisomy 21 is the most common chromosome abnormality, occurring in 1 in 700 to 800 live births. This condition usually happens by chance and is associated with increasing 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 trisomy 21.
- 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 trisomy 21, but a normal ultrasound cannot exclude this condition.

Resources for trisomy 21:

Genetics Home Reference http://ghr.nlm.nih.gov/chromosome/21

National Down Syndrome Society (NDSS) - http://www.ndss.org

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:

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Malvestiti F, Agrai C, Grimi B, et al. Interpreting mosaicism in chorionic villi: results of a monocentric series of 1001 mosaics in chorionic villi with follow-up amniocentesis. *Prenat Diagn.* 2015;35:1117-1127.

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