General information about positive NIPT results

My patient's NIPT is positive for 13 (Patau syndrome). What does this mean? Your patient's NIPT result suggests the presence of an extra copy of chromosome 13. NIPT is a screening test; false positives can occur. The actual chance for the pregnancy to have trisomy 13 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 13 and additional resources.

What is trisomy 13? Trisomy 13 is a condition that is caused by an extra chromosome number 13 (three copies instead of two).

What are the features of trisomy 13? Although the majority of pregnancies with trisomy 13 result in miscarriage or stillbirth, trisomy 13 can result in live birth. Individuals with trisomy 13 have severe intellectual disability and abnormalities involving multiple organs. Some of the common features of trisomy 13 include heart defects, omphalocele, brain abnormalities such as holoprosencephaly, cleft lip and palate, and other features. Although less than 10% of babies with trisomy 13 will live past 1 year of age, some people with this condition can live years or even decades.

What is the prevalence of this condition? Trisomy 13 occurs in approximately 1 in 12,000 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 13.
- 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 13, but a normal ultrasound cannot exclude this condition.

Resources for trisomy 13:

Genetics Home Reference http://ghr.nlm.nih.gov/chromosome/13

Unique, The Rare Chromosome Disorder Support Group http://www.rarechromo.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:

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.

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.