General information about positive NIPT results

My patient's NIPT is positive for 1p36 deletion syndrome. What does this mean? Your patient's screening test detected a deletion of 1p36 which is associated with 1p36 deletion syndrome. NIPT is a screening test; false positives can occur.

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 the general description of 1p36 deletion syndrome and additional resources.

What is 1p36 deletion syndrome? 1p36 deletion syndrome is a genetic syndrome characterized by birth defects, intellectual disability, and other serious medical issues. 1p36 deletion syndrome is caused by a deletion in the region of 1p36.

What are the common features of 1p36 deletion syndrome? Key features of this syndrome include: characteristic craniofacial features, intellectual disability, seizures, skeletal abnormalities, and brain and heart defects. Lifespan is variable, but can be normal.

What is the prevalence of this condition? 1 in 4,000-10,000 newborns have this condition, with a female: male ratio of 2:1.

What testing could be considered?

- Specialized genetic tests such as fluorescence in situ hybridization (FISH) and microarray are available to confirm the presence of 1p36 deletion 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 in case of a miscarriage.
- Ultrasound evaluation may be useful in aiding with a prenatal diagnosis of 1p36 deletion syndrome, but a normal ultrasound does not exclude the presence of the condition.

Resources for 1p36 deletion syndrome:

1p36 Deletion Support & Awareness
http://www.1p36dsa.org/what-is-1p36-deletion-syndrome/

1p36 deletion syndrome-Genetics Home Reference
http://ghr.nlm.nih.gov/condition/1p36-deletion-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:

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

Giannikou K, Fryssira H, Oikonomakis V, et al. Further delineation of novel 1p36 rearrangements by array-CGH analysis: narrowing the breakpoints and clarifying the "extended" phenotype. *Gene.* 2012;506:360-368.

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