General information about positive NIPT results

My patient's NIPT is positive for Cri-du-Chat (5p-) syndrome. What does this mean? Your patient's screening test detected a deletion of 5p15.2 which is associated with 5p- syndrome (also known as Cri-du-Chat 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 5p- syndrome and additional resources.

What is 5p- syndrome? 5p- syndrome is a genetic syndrome characterized by birth defects, intellectual disability, and other serious medical issues. 5p- syndrome is caused by a deletion in the region of 5p15.2.

What are the features of 5p- syndrome? Key features of this syndrome include: significant intellectual disability, speech delay, cat-like cry, dysmorphic features, microcephaly and 10% mortality in first year.

What is the prevalence of this condition? 1 in 20,000-50, 000 live births have this condition.

What testing could be considered?

- Specialized genetic tests such as fluorescence in situ hybridization (FISH) and microarray are available to confirm the presence of 5p- 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 5p- syndrome, but a normal ultrasound does not exclude the presence of the condition.

Resources for 5p- syndrome:

5p- Society http://www.fivepminus.org/

Cri-du-Chat syndrome- National Organization for Rare Disorders https://rarediseases.org/rare-diseases/cri-du-chat-syndrome/

Cri-du-Chat syndrome-Genetics Home Reference http://ghr.nlm.nih.gov/condition/cri-du-chat-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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