General information about positive NIPT results

My patient's NIPT is positive for a deletion of 22q11.2.
What does this mean? Your patient's screening test detected a deletion of 22q11.2 which is associated with 22q11.2 deletion syndrome (also known as DiGeorge syndrome or Velocardiofacial 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 22q11.2 deletion syndrome and additional resources.

What is 22q11.2 deletion syndrome? 22q11.2 deletion syndrome is a genetic syndrome that is variable in presentation. Many features have been reported, yet individuals with this syndrome may have different presentations from one another.

What are the features of 22q11.2 deletion syndrome? Key features of this syndrome are variable, but include: intellectual disability, heart defects, palatal abnormalities, immune deficiency, and dysmorphic features. Life span is usually normal, but can vary depending on severity of features.

What is the prevalence of this condition? 1 in 4,000 live births have this condition.

What testing could be considered?

- NIPT can only screen for cases of 22q11.2 deletion syndrome caused by the common 3MB deletion, which accounts for 85% of patients with DiGeorge/velocardiofacial syndrome.
- Specialized genetic tests such as fluorescence in situ hybridization (FISH) and microarray are available to confirm the presence of 22q11.2 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 (POC) in case of a miscarriage.
- Ultrasound evaluation may be useful in aiding with a prenatal diagnosis of 22q11.2 deletion syndrome, but a normal ultrasound cannot exclude this condition.

Resources for 22q11.2 deletion syndrome:

The International 22q11.2 Foundation Inc. http://www.22q.org/

22q11.2 deletion syndrome-Genetics Home Reference
http://ghr.nlm.nih.gov/condition/22q112-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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Jones KL. *Smith's Recognizable Patterns of Human Malformation.* 5th ed. Philadelphia, PA: W.B. Saunders Company; 1997.

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