General information about positive NIPT results: Trisomy 19

My patient's NIPT is positive for trisomy 19. What does this mean?

Your patient's noninvasive prenatal testing (NIPT) result suggests the presence of an extra copy of chromosome 19. NIPT is a screening test; false positives can occur. The actual chance for the pregnancy to have trisomy 19 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. Globally, professional medical societies recommend that all women with a positive screening result should have genetic counseling and a comprehensive ultrasound evaluation with an opportunity for diagnostic testing to confirm the results.[1,2] Confirmation prior to birth can also help with pregnancy and neonatal management.

See below for more information about trisomy 19.

What is trisomy 19?

Trisomy 19 is a condition that is caused by an extra chromosome number 19 (three copies instead of two).

What are the features of trisomy 19?

Full trisomy 19 has not been reported in live births and presumably leads to early pregnancy loss. Few cases of mosaic trisomy 19 (where some cells are normal and some cells have trisomy 19) have been described in the literature. These cases are expected to have a wide range of medical complications, as well as physical and developmental sequelae. Prenatal ultrasound may be normal. Confined placental mosaicism (CPM; when trisomic cells are present in the placenta, but not in the fetus) has not been frequently reported for trisomy 19.

What is the prevalence of this condition?

Unknown, but extremely rare. For this reason, positive predictive value (PPV) cannot be accurately calculated.

What testing could be considered?

Specialized genetic tests such as karyotyping, fluorescence *in situ* hybridization (FISH), quantitative polymerase chain reaction (qPCR), and microarray are available to confirm the presence of trisomy 19.

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 the case of a miscarriage. The type of invasive procedure and diagnostic testing should take into account the underlying chromosome anomaly.[3,4]

Ultrasound evaluation may be useful in aiding with a prenatal diagnosis of trisomy 19, but a normal ultrasound cannot exclude this condition.

Resources for trisomy 19

MedlinePlus Genetics
medlineplus.gov/genetics/chromosome/19

Unique, The Rare Chromosome Disorder Support Group
rarechromo.org

References

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Additional resources

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