General information about positive NIPT results: Trisomy 17

My patient's NIPT is positive for Trisomy 17. What does this mean?

Your patient's noninvasive prenatal testing (NIPT) result suggests the presence of an extra copy of chromosome 17. NIPT is a screening test; false positives can occur. The actual chance for the pregnancy to have trisomy 17 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 17.

What is trisomy 17?

Trisomy 17 is a condition that is caused by an extra chromosome number 17 (three copies instead of two).

What are the features of trisomy 17?

Most pregnancies with trisomy 17 will miscarry spontaneously. Full trisomy 17 has not been reported in live births and presumably leads to early pregnancy loss. If a developing fetus has mosaic trisomy 17 (where some cells are normal and some cells have trisomy 17), there is an increased chance for the pregnancy to progress and possibly survive to term. However, liveborn infants with mosaic trisomy 17 are expected to have serious medical problems. Key features include intellectual disability, growth restriction, and organ system anomalies. In reported cases of prenatally diagnosed trisomy 17, the outcomes have ranged from normal to live births with clinical sequelae. The variability in clinical presentation is believed to be due to confined placental mosaicism (CPM; when trisomic cells are present in the placenta, but not in the fetus) or the degree of fetal mosaicism.

What is the prevelance of this condition?

Unknown, but 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 17.

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 17, but a normal ultrasound cannot exclude trisomy 17.

Resources for trisomy 17

MedlinePlus Genetics
medlineplus.gov/genetics/chromosome/17

Unique, The Rare Chromosome Disorder Support Group
rarechromo.org

References

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Additional resources

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