General information about positive NIPT results: Trisomy 12

My patient's NIPT is positive for trisomy 12. What does this mean?

Your patient's noninvasive prenatal testing (NIPT) result suggests the presence of an extra copy of chromosome 12. NIPT is a screening test; false positives can occur. The actual chance for the pregnancy to have trisomy 12 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 12.

What is trisomy 12?

Trisomy 12 is a condition that is caused by an extra chromosome number 12 (three copies instead of two).

What are the features of trisomy 12?

Most pregnancies with trisomy 12 will miscarry spontaneously. Full trisomy 12 has not been reported in live births. If a developing fetus has mosaic trisomy 12 (where some cells are normal and some cells have trisomy 12), there is an increased chance for the pregnancy to progress and possibly survive to term. However, liveborn infants with mosaic trisomy 12 are expected to have serious medical problems. Key features include intellectual disability, dysmorphism, and organ system anomalies, and can lead to fetal or neonatal death. 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 prevalence 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 12.

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 12, but a normal ultrasound cannot exclude trisomy 12.

Resources for trisomy 12

MedlinePlus Genetics
medlineplus.gov/genetics/chromosome/12

Unique, The Rare Chromosome Disorder Support Group
rarechromo.org

References

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Additional resources

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