General information about positive NIPT results: Trisomy 1

My patient's NIPT is positive for trisomy 1. What does this mean?

Your patient's noninvasive prenatal testing (NIPT) result suggests the presence of an extra copy of chromosome 1. NIPT is a screening test; false positives can occur. The actual chance for the pregnancy to have trisomy 1 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 1.

What is trisomy 1?

Trisomy 1 is a condition that is caused by an extra chromosome number 1 (three copies instead of two).

What are the features of trisomy 1?

Most pregnancies with trisomy 1 will miscarry spontaneously. All reported cases of prenatally diagnosed trisomy 1 have resulted in blighted ovum and no cases of full trisomy 1 have been reported in a live birth. If a developing fetus has mosaic trisomy 1 (where some cells are normal and some cells have trisomy 1), there is an increased chance for the pregnancy to progress and possibly survive to term. However, liveborn infants with mosaic trisomy 1 are extremely rare and are expected to have a wide range of medical complications and physical and developmental sequelae, not all of which may be detected by prenatal ultrasound. Confined placental mosaicism (CPM; when trisomic cells are present in the placenta, but not in the fetus) has not been frequently reported for trisomy 1.

What is the prevalence of this condition?

Unknown, but very rare. For this reason, positive predictive value (PPV) cannot be 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 1.

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 1, but a normal ultrasound cannot exclude this condition.

Resources for trisomy 1

MedlinePlus Genetics
medlineplus.gov/genetics/chromosome/1

The Rare Chromosome Disorder Support Group
rarechromo.org

References

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Additional resources

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