General information about positive NIPT results: Trisomy 11

My patient's NIPT is positive for trisomy 11. What does this mean?

Your patient's noninvasive prenatal testing (NIPT) result suggests the presence of an extra copy of chromosome 11. NIPT is a screening test; false positives can occur. The actual chance for the pregnancy to have trisomy 11 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 11.

What is trisomy 11?

Trisomy 11 is a condition that is caused by an extra chromosome number 11 (three copies instead of two).

What are the features of trisomy 11?

Full trisomy 11 has not been reported in live births and presumably leads to early pregnancy loss. Almost all reported live births with a prenatal diagnosis of mosaic trisomy 11 have normal prenatal and postnatal outcome, without evidence of trisomy 11 postnatally.

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), qPCR, and microarray are available to confirm the presence of trisomy 11. Uniparental disomy (UPD) analysis is performed by specialized testing, such as single nucelotide polymorphism (SNP) microarray, methylation testing, and short tandem repeat (STR) marker testing.

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 11, but a normal ultrasound cannot exclude this condition.

Special considerations

Chromosome 11 is an imprinted chromosome and UPD for chromosome 11 has specific consequences. There may be an increased risk for certain recessive conditions if UPD is present. Paternal UPD 11 is associated with Beckwith-Wiedemann syndrome. Maternal UPD 11 may be associated with Russell-Silver syndrome.[5]

The American College of Medical Genetics and Genomics (ACMG) states that specialized UPD testing should be considered for patients when there is discordance noted between the NIPT result and diagnostic testing.[4]

Resources for trisomy 11

MedlinePlus Genetics
medlineplus.gov/genetics/chromosome/11

Unique, The Rare Chromosome Disorder Support Group
rarechromo.org

References

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Additional resources

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