General information about positive NIPT results: Trisomy 20

My patient's NIPT is positive for trisomy 20. What does this mean?

Your patient's noninvasive prenatal testing (NIPT) result suggests the presence of an extra copy of chromosome 20. NIPT is a screening test; false positives can occur. The actual chance for the pregnancy to have trisomy 20 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 20.

What is trisomy 20?

Trisomy 20 is a condition that is caused by an extra chromosome number 20 (three copies instead of two).

What are the features of trisomy 20?

Most pregnancies with trisomy 20 will miscarry spontaneously. If a developing fetus has mosaic trisomy 20 (where some cells are normal and some cells have trisomy 20), there is an increased chance for the pregnancy to progress and possibly survive to term. However, liveborn infants with full or mosaic trisomy 20 are expected to have serious medical problems. Full trisomy 20 is rarely reported in live births and is associated with anomalies often involving the central nervous system and heart. Mosaic trisomy 20 is associated with an increased risk of organ system anomalies (most commonly cardiac and renal anomalies), growth restriction, intellectual disability, and dysmorphism. In reported cases of prenatally diagnosed trisomy 20, 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. Mosaic trisomy 20 is detected in 1 in 5000 amniocentesis samples.[3]

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 20. Uniparental disomy (UPD) analysis is performed by specialized testing, such as single nucleotide 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.[4,5]

Ultrasound evaluation may be useful in aiding with a prenatal diagnosis of trisomy 20, but a normal ultrasound cannot exclude this condition.

Special considerations

Chromosome 20 is an imprinted chromosome and UPD for chromosome 20 has specific consequences. Maternal UPD 20 is associated with poor growth (in utero and postnatally) and feeding difficulties. Paternal UPD 20 is associated with pseudohypoparathyroidism type 1B. There may be an increased risk for certain recessive conditions if UPD is present.

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 NIPT results and the diagnostic testing.[6]

Resources for trisomy 20

MedlinePlus Genetics
medlineplus.gov/genetics/chromosome/20

Unique, The Rare Chromosome Disorder Support Group
rarechromo.org

References

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Additional resources

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