General information about positive NIPT results: Trisomy 22

My patient's NIPT is positive for trisomy 22. What does this mean?

Your patient's noninvasive prenatal testing (NIPT) result suggests the presence of an extra copy of chromosome 22. NIPT is a screening test; false positives can occur. The actual chance for the pregnancy to have trisomy 22 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 22.

What is trisomy 22?

Trisomy 22 is a condition that is caused by an extra chromosome number 22 (three copies instead of two).

What are the features of trisomy 22?

Most pregnancies with trisomy 22 will miscarry spontaneously. If a developing fetus has mosaic trisomy 22 (where some cells are normal and some cells have trisomy 22), there is an increased chance for the pregnancy to progress and possibly survive to term. However, liveborn infants with full or mosaic trisomy 22 are expected to have serious medical problems. Key features include: intrauterine growth restriction, dysmorphism, webbed neck, and organ system anomalies including cardiac and renal anomalies. In reported cases of prenatally diagnosed trisomy 22, 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. Full trisomy 22 is rarely reported in live births and is usually associated with a poor, typically fatal prognosis. Most liveborn infants with mosaic trisomy 22 do not survive past the first few months. When mosaic trisomy 22 is detected by amniocentesis, the risk for abnormal outcome is > 60%.[3,4]

What is the prevalence of this condition?

Full trisomy 22 is rare and may be about 1 in 30,000 to 50,000 live births.[5] Since the prevalence is not clearly known, 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 22.

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.[6,7]

Ultrasound evaluation may be useful in aiding with a prenatal diagnosis of trisomy 22, but a normal ultrasound cannot exclude this condition.

Resources for trisomy 22

MedlinePlus Genetics
medlineplus.gov/genetics/chromosome/22

Unique, The Rare Chromosome Disorder Support Group
rarechromo.org

References

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