General information about positive NIPT results: Trisomy 2

My patient's NIPT is positive for trisomy 2. What does this mean?

Your patient's noninvasive prenatal testing (NIPT) result suggests the presence of an extra copy of chromosome 2. NIPT is a screening test; false positives can occur. The actual chance for the pregnancy to have trisomy 2 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 2.

What is trisomy 2?

Trisomy 2 is a condition that is caused by an extra chromosome number 2 (three copies instead of two).

What are the features of trisomy 2?

Most pregnancies with trisomy 2 will miscarry spontaneously. If a developing fetus has mosaic trisomy 2 (where some cells are normal and some cells have trisomy 2), there is an increased chance for the pregnancy to progress and possibly survive to term. However, liveborn infants with full or mosaic trisomy 2 are expected to have serious medical problems. Key features include: growth and motor delay, intellectual disability and congenital anomalies such as microcephaly, cleft lip and palate, congenital heart defects, etc. In reported cases of prenatally diagnosed trisomy 2, 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. Ultrasound may reveal intrauterine growth restriction, oligohydramnios, or other anomalies.

What is the prevalence of this condition?

Trisomy 2 has been reported in 1 in 2000 chorionic villus sampling (CVS) results[3,4] and 1 in 58,000 amniocentesis results.[5] The exact prevalence is unknown, therefore 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 2.

These confirmatory tests are generally performed on cells from 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 2, but a normal ultrasound cannot exclude this condition.

Resources for trisomy 2

MedlinePlus Genetics
medlineplus.gov/genetics/chromosome/2

Unique, The Rare Chromosome Disorder Support Group
rarechromo.org

References

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Additional resources

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