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General information about positive NIPT results: Trisomy 13 (Patau syndrome)

My patient's NIPT is positive for Trisomy 13 (Patau syndrome). What does this mean?

Your patient's noninvasive prenatal testing (NIPT) result suggests the presence of an extra copy of chromosome 13. NIPT is a screening test; false positives can occur. The actual chance for the pregnancy to have trisomy 13 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 13.

What is trisomy 13?

Trisomy 13 is a condition that is caused by an extra chromosome number 13 (three copies instead of two).

What are the features of trisomy 13?

Although the majority of pregnancies with trisomy 13 result in miscarriage or stillbirth, trisomy 13 can result in live birth. Individuals with trisomy 13 have severe intellectual disability and abnormalities involving multiple organs. Some of the common features of trisomy 13 include heart defects, omphalocele, brain abnormalities such as holoprosencephaly, cleft lip and palate, and other features. Although less than 10% of babies with trisomy 13 will live past 1 year of age, some people with this condition can live years or even decades.

What is the prevelance of this condition?

Trisomy 13 occurs in approximately 1 in 12,000 live births. This condition usually happens by chance and is associated with increasing maternal age.

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 13.

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 13, but a normal ultrasound cannot exclude trisomy 13.

Resources for trisomy 13

MedlinePlus Genetics
medlineplus.gov/genetics/chromosome/13

Unique, The Rare Chromosome Disorder Support Group
rarechromo.org

References

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Additional resources

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Malvestiti F, Agrati C, Grimi B, et al. Interpreting mosaicism in chorionic villi: results of a monocentric series of 1001 mosaics in chorionic villi with follow-up amniocentesis. *Prenat Diagn*. 2015;35(11):1117-1127.

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