General information about positive NIPT results: Trisomy 18 (Edwards syndrome)

My patient's NIPT is positive for Trisomy 18 (Edwards syndrome). What does this mean?

Your patient's noninvasive prenatal testing (NIPT) result suggests the presence of an extra copy of chromosome 18. NIPT is a screening test; false positives can occur. The actual chance for the pregnancy to have trisomy 18 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 18.

What is trisomy 18?

Trisomy 18 is a condition that is caused by an extra chromosome number 18 (three copies instead of two).

What are the features of trisomy 18?

Although many pregnancies with trisomy 18 result in miscarriage or stillbirth, trisomy 18 can result in livebirth. Individuals with trisomy 18 have severe intellectual disability and abnormalities involving multiple organs. Some of the common features of trisomy 18 include heart defects, brain abnormalities, musculoskeletal problems, cleft lip and palate, clenched hand, and low birth weight. Although less than 10% of babies with trisomy 18 will live past 1 year of age, some people with this condition can live years or even decades.

What is the prevalence of this condition?

Trisomy 18 occurs in 1 in 6000 to 8000 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 18.

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 18, but a normal ultrasound cannot exclude trisomy 18.

Resources for trisomy 18

MedlinePlus Genetics
medlineplus.gov/genetics/chromosome/18

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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