General information about positive NIPT results: Monosomy X (Turner syndrome)

My patient's NIPT is positive for monosomy X (Turner syndrome). What does this mean?

Your patient's noninvasive prenatal testing (NIPT) result suggests the presence of one X sex chromosome and the absence of a second sex chromosome. NIPT is a screening test; false positives can occur. The actual chance for the pregnancy to have monosomy X 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 monosomy X.

What is monosomy X?

Monosomy X is a condition that is caused by having one X chromosome and an absent second sex chromosome (one sex chromosome instead of two sex chromosomes).

What are the features of monosomy X?

Many pregnancies with monosomy X will result in a pregnancy loss; however, monosomy X is compatible with continued survival and live birth. Females with monosomy X have variable phenotypes. Typically, females with monosomy X have normal intelligence; however, learning disabilities are possible and variable. Some of the common features of monosomy X include heart defects, kidney abnormalities, cystic hygroma, short stature, congenital lymphedema, and primary ovarian failure.

What is the prevalence of this condition?

Approximately 1 in 2500 females are born with monosomy X. This condition usually happens by chance and is not typically associated with advanced parental 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 monosomy X.

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 monosomy X, but a normal ultrasound cannot exclude this condition.

Resources for monosomy X

Turner Syndrome Society of the United States
turnersyndrome.org

Turner Syndrome Support Society of the United Kingdom
tss.org.uk

References

1. Rose NC, Kaimal AJ, Dugoff L, et al. Screening for Fetal Chromosomal Abnormalities: ACOG Practice Bulletin, Number 226. *Obstet Gynecol*. 2020;136(4):e48-e69.
2. Benn P, Borrell A, Chiu RW, et al. Position statement from the Chromosome Abnormality Screening Committee on behalf of the Board of the International Society for Prenatal Diagnosis. *Prenat Diagn*. 2015;35(8):725-734.
3. Cherry AM, Akkari YM, Barr KM, et al. Diagnostic cytogenetic testing following positive noninvasive prenatal screening results: a clinical laboratory practice resource of the American College of Medical Genetics and Genomics (ACMG). *Genet Med*. 2017;19(8):845-850.
4. Van Opstal D, Srebniak MI. Cytogenetic confirmation of a positive NIPT result: evidence-based choice between chorionic villus sampling and amniocentesis depending on chromosome aberration. *Expert Rev Mol Diagn*. 2016;16(5):513-520.

Additional resources

Gardner RJM, Sutherland GR, Shaffer LG. *Gardner and Sutherland's Chromosome Abnormalities and Genetic Counseling*. 5th ed. Oxford University Press; 2018.

Jones KL, Jones MC, del Campo M. *Smith's Recognizable Patterns of Human Malformation*. 7th ed. W.B. Saunders Company; 2013.

Pub. No. 1576-2020-032 QB9516