Triple X Syndrome

Triple X syndrome is a sex chromosome aneuploidy that occurs in females when there is an extra copy of the X chromosome meaning there are three copies instead of the expected two (figure 1). It is a chromosomal condition occurring in at least 1 in every 1000 female births, however it is estimated that only 10% of females with Triple X syndrome ever come to clinical attention so it could be even more common. Some females with Triple X syndrome will be mosaic, where some of their cells have two X chromosomes and the other cells have three X chromosomes.

Babies that are born with Triple X syndrome could have a number of the features and symptoms, however, not everyone will have them all and severity will vary significantly. Mosaicism plays a role in the varied features and severity of Triple X syndrome. Some features and symptoms include learning difficulties, motor and speech delay, psychological conditions and tall stature. Some females with Triple X syndrome will not have any features. Although there is no cure for Triple X syndrome evidence suggests that early intervention services and treatment can assist in some of the associated symptoms if they present.

What does a Non-invasive Prenatal Test (NIPT) result mean?

Receiving a high probability result for Triple X syndrome means that there is a significant chance that a baby will have Triple X syndrome. However, NIPT is a screening test meaning, in some rare instances, the baby may not actually have the condition. There are many different reasons for these results to be different and some of them cannot be avoided. Due to this, a high probability NIPT result should be confirmed using a diagnostic test before making any major clinical decisions. These tests involve an invasive procedure, known as Chorionic Villus Sampling (CVS) or amniocentesis. A doctor, midwife or genetic counsellor can provide information on the different options available.

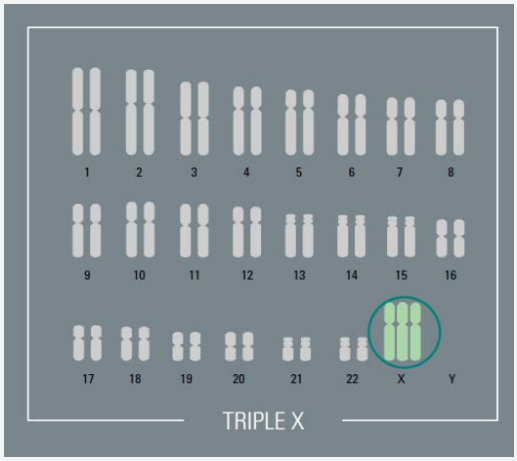

Figure 1 Karyotype showing a person with three copies of the X chromosome. This person would have Triple X syndrome.

It is important for each individual or family to understand what these results mean for them and their situation. Your healthcare provider should be able to answer a number of questions you may have about the condition and your results. Further information can be found by visiting the references below.

This information sheet provides general information about the test result and recommendations. It is not intended to replace genetic counselling by a medical professional.

Non-invasive prenatal testing (NIPT) based on cell-free analysis is not diagnostic: results should be confirmed by diagnostic testing. Before making any treatment decisions, all women should discuss their results with their healthcare provider, who can recommend confirmatory, diagnostic testing where appropriate

Reference Material:

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