Monosomy X – Turner Syndrome

Turner syndrome, or Monosomy X, is a sex chromosome aneuploidy that occurs in females when there is only one copy of the X chromosome instead of the expected two (figure 1). It occurs in at least 1 in every 2500 female births. Unfortunately, many pregnancies with Turner syndrome will miscarry in the first or second trimester. More than half of those with Turner syndrome will be mosaic, where some of their cells have just one X chromosome and the other cells have two X chromosomes.

Babies that are born with Turner 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 severity of Turner syndrome. Features and symptoms include subtle changes in physical appearance, short stature, infertility and learning difficulties as well as some potential health conditions including cardiac conditions, hypothyroidism, diabetes and autoimmune disease. Although there is no cure for Turner syndrome many of the associated symptoms can be treated. Girls with Turner syndrome will have regular health checks of their heart, kidneys and reproductive system throughout their lives and will usually lead a relatively normal and healthy life.

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

Receiving a high probability result for Turner syndrome means that there is a significant chance that a baby will have Turner 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.

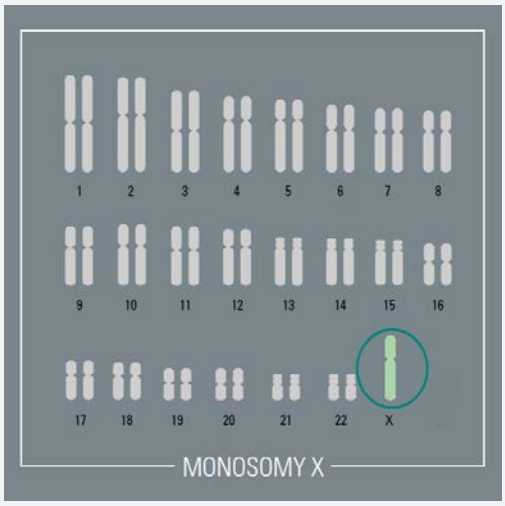

Figure 1 Karyotype showing a female with one copy of the X chromosome. This person would have Turner syndrome.

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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