47,XXY - Klinefelter Syndrome

Klinefelter syndrome is a sex chromosome aneuploidy that occurs in males when there are two copies of the X chromosome instead of the expected one X chromosome (figure 1). It is a common chromosomal condition occurring in at least 1 in every 1000 male births, possibly as many as 1 in 500. Many males with Klinefelter syndrome may never be diagnosed. Some males with Klinefelter syndrome may be mosaic, meaning some of their cells have two X and one Y chromosome and the other cells have one X and one Y chromosome.

Babies that are born with Klinefelter 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 Klinefelter syndrome. Features and symptoms include learning difficulties, developmental delay, infertility as well as some physical features. While there is no cure for Klinefelter syndrome, many of the problems can usually be treated if they do occur and testosterone replacement therapy may help reduce the risk of some symptoms.

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

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

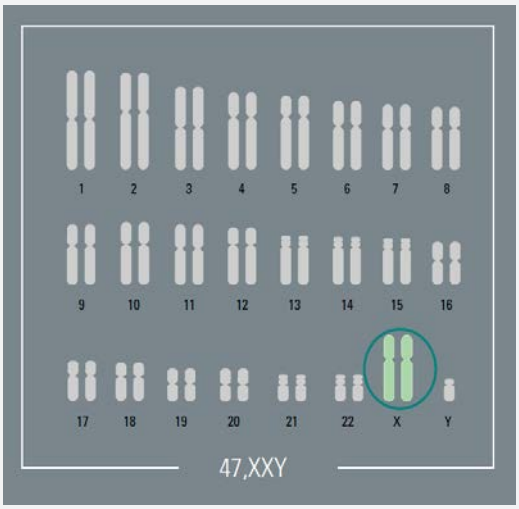

Figure 1 Karyotype showing a male with two copies of the X chromosome. This person would have Klinefelter syndrome.

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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