XYY Syndrome – Jacobs Syndrome

XYY syndrome, sometimes known as Jacobs syndrome, is a sex chromosome aneuploidy that occurs in males when there are two copies of the Y chromosome instead of the expected one Y chromosome (figure 1). It is a chromosomal condition occurring in at least 1 in every 1000 male births; however, many males with XYY syndrome never come to clinical attention so it could be more common. Some males with XYY syndrome will be mosaic, where some of their cells have two Y chromosomes and the other cells have the usual one Y chromosome.

Many babies that are born with XYY syndrome do not have any clinical features and symptoms. There are no fertility issues associated with XYY syndrome. Males with XYY syndrome can be very tall and may have severe acne during adolescence. The average intellectual capacity of males with XYY syndrome is within the normal range; however their IQ may be slightly lower than their siblings. Learning difficulties have been associated in some people with XYY syndrome, usually involving speech and language. There are many misconceptions about males with XYY syndrome, previously, it was sometimes called the super-male disease and was associated with being overly-aggressive and lacking in empathy. Recent studies have disproven this and they are no longer associated with XYY syndrome.

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

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

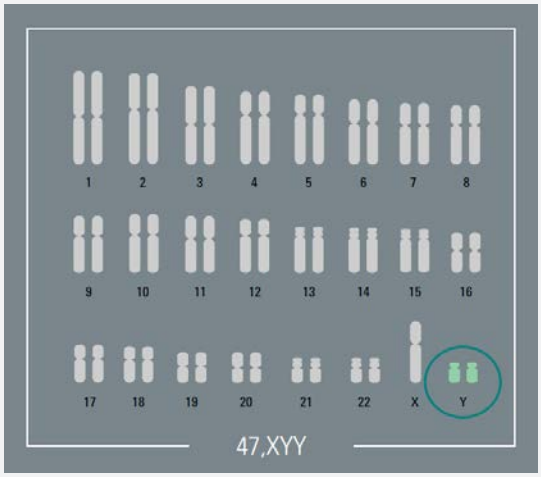

Figure 1 Karyotype showing a male with two copies of the Y chromosome. This person would have XYY syndrome.

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,

Reference Material

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