Sonic Genetics
Quality is in our DNA

harmony®
PRENATAL TEST

XYY / Jacobs syndrome

Information for patients

The healthcare professional responsible for your care has given you this leaflet because you have been identified by the Harmony® Prenatal Test as having a high probability of a chromosome disorder in your pregnancy. This fact sheet contains more information about the particular genetic disorder mentioned in your Harmony report. We recommend that you also discuss your result with an experienced doctor or genetic counsellor.

XYY syndrome, sometimes known as Jacobs syndrome, is a sex chromosome disorder 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 one in every 1,000 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, meaning some of their cells have two Y chromosomes and the other cells have the usual one Y chromosome.

Males with XYY syndrome can be very tall and may have severe acne during adolescence. The average intellectual capacity is within the normal range, although 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 no fertility issues associated with the syndrome, and many babies that are born with it do not have any clinical features and symptoms at all. 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 disproved this, and these traits are no longer associated with the syndrome.

How to interpret a high probability result for XYY syndrome

Receiving a high probability result for XYY syndrome means that there is an increased chance that your baby will have XYY syndrome. However, it is important to remember that all NIPTs are screening tests, and for many different biological reasons the baby may not have the condition that is reported as high probability. 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.

It is important for each individual or family to understand what these results mean for them and their situation. Your healthcare provider is the person to ask if you have any questions about the condition and your results. Further information can be found by visiting the references listed below.

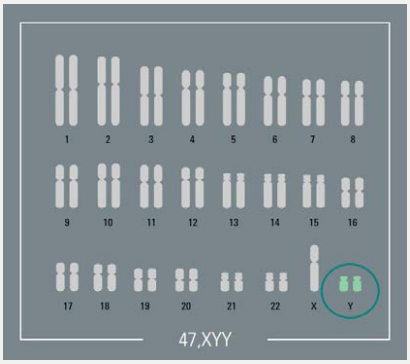

Figure 1 Karyotype showing a male with two copies of the Y chromosome. This person would have XYY syndrome.

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

References

- National Library of Medicine (US). Genetics Home Reference [Internet]. Bethesda (MD): The Library; 2017 Nov 21. XYY syndrome; [reviewed 2009 Jan]
- National Organisation for Rare Disorders. XYY syndrome [Internet]. 2012
- National Institute of Health (US). Genetic and Rare Diseases [Internet]. Gaithersburg (MD). 47, XYY syndrome; Oct 16 2017
- Unique. The Rare Chromosome Disorder Support Group [Internet]. Disorder Guides. XYY. 2014

For further information, including scientific and peer-reviewed publications, please refer to our website, www.sonicgenetics.com.au/nipt or call us on 1800 010 447

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