Sonic Genetics
Quality is in our DNA

harmony®
PRENATAL TEST

47,XXY Klinefelter 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.

Klinefelter syndrome is a sex chromosome disorder 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 one in every 1,000 male births, possibly as many as one 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.

The features and symptoms of Klinefelter syndrome include learning difficulties, developmental delay and infertility, as well as some physical features. Babies who are born with the syndrome could have a number of the features and symptoms, however, not everyone will have them all and severity will vary significantly. Mosaicism also plays a role in the varied features and severity of the syndrome. 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 to reduce the severity of some symptoms.

How to interpret a high probability result for Klinefelter syndrome

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

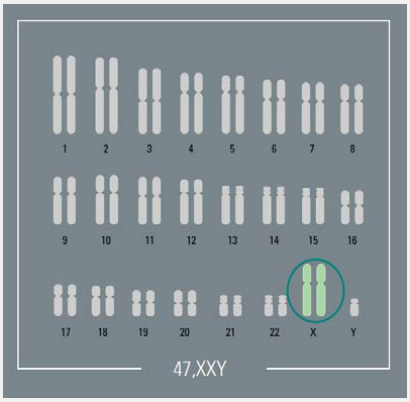

Figure 1 Karyotype showing a male with two copies of the X chromosome. This person would have Klinefelter syndrome.

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

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- National Library of Medicine (US). Genetics Home Reference [Internet]. Bethesda (MD): The Library; 2017 Nov 14. Klinefelter syndrome; [reviewed 2013 Jan]
- Visootsak J, Graham JM. *Orphanet J Rare Dis.* 2006;1:42

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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