Sonic Genetics
Quality is in our DNA

harmony®
PRENATAL TEST

Monosomy X
Turner 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.

Turner syndrome, or Monosomy X, is a sex chromosome disorder 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 one in every 2,500 female births. Monosomy X may be associated with an increased risk of miscarriage in the first or second trimester. More than half of those with Turner syndrome will be mosaic, meaning some of their cells have just one X chromosome and the other cells have two X chromosomes.

Features and symptoms of Turner syndrome 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. Babies who are born with Turner syndrome could have a number of the features and symptoms of the syndrome, however, not everyone will have them all and severity will vary significantly. Mosaicism also plays a role in the varied severity of the syndrome. Although there is no cure for Turner syndrome, many of the associated symptoms can be treated. Girls with Turner syndrome may need regular health checks of their heart, kidneys and reproductive system throughout their lives.

How to interpret a high probability result for Monosomy X

Receiving a high probability result for Turner syndrome means that there is an increased chance that your baby will have Turner 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 about this condition can be found by visiting the references listed below.

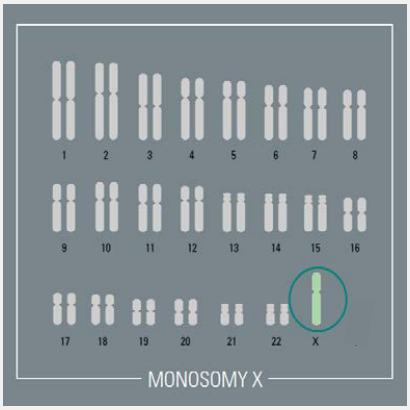

Figure 1 Karyotype showing a female with one copy of the X chromosome. This person would have Turner syndrome.

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

- A.D.A.M. Medical Encyclopedia. Atlanta (GA): A.D.A.M., Inc.; ©2005. Turner syndrome;2015 Oct 27
- Centre for Genetics Education (AU). Fact sheet 40 - Turner syndrome [Internet]. 2015 Nov
- National Library of Medicine (US). Genetics Home Reference [Internet]. Bethesda (MD): The Library; 2017 Nov 21. Turner syndrome; [reviewed 2014 Jun]
- National Organization for Rare Disorders. Turner syndrome [Internet]. 2012
- Mayo Clinic for Medical Education and Research. Turner Syndrome. August 20, 2011

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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