Sonic Genetics
Quality is in our DNA

harmony®
PRENATAL TEST

Triple X 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.

Triple X syndrome is a sex chromosome disorder that occurs in females when there is an extra copy of the X chromosome, meaning there are three copies instead of the expected two (Figure 1). It is a chromosomal condition occurring in at least one in every 1,000 female births, however, it is estimated that only 10% of females with Triple X syndrome ever come to clinical attention, so it could be even more common. Some females with Triple X syndrome will be mosaic, meaning some of their cells have two X chromosomes and the other cells have three X chromosomes.

Some features and symptoms of Triple X syndrome include learning difficulties, motor and speech delay, psychological conditions and tall stature. 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. Some females with the syndrome will not have any features at all. Mosaicism also plays a role in the varied features and severity of the syndrome. Although there is no cure for Triple X syndrome, evidence suggests that early intervention services and treatment can assist in some of the associated symptoms if they present.

How to interpret a high probability result for Triple X syndrome

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

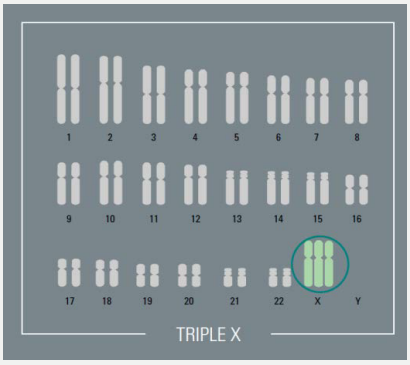

Figure 1 Karyotype showing a person with three copies of the X chromosome. This person would have Triple X syndrome.

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 14. Triple X syndrome; [reviewed 2013 Jan]
- Otter et al. *Eur J Hum Genet.* 2010 Mar; 18(3): 265–271
- Tartaglia et al. *Orphanet J Rare Dis.* May 2010; 11:58
- Samango-Sprouse C. National Organisation for Rare Disorders (NORD). 2014
- Triple X syndrome. Mayo Clinic. December, 2015

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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