Sonic Genetics
Quality is in our DNA

harmony® PRENATAL TEST

Trisomy 18 Edwards 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.

Edwards syndrome, otherwise known as Trisomy 18, is a serious chromosomal condition that occurs when there is an extra copy of chromosome 18, meaning that there are three copies instead of the expected two (Figure 1). Edwards syndrome occurs in about one in 5,000 newborns and most babies with Edwards syndrome will miscarry; of those babies who are born with Edwards syndrome, most will live for just a short time. While the chance of having a baby with Edwards syndrome increases as women age, it can occur in any pregnancy.

Edwards syndrome can affect many different parts of the body, including abnormalities of the heart, brain, kidneys and gastro-intestinal system. Edwards syndrome can also cause failure to grow, development delay, intellectual disability and other physical features. There is no cure for Edwards syndrome and the symptoms can be difficult to manage. You are likely to need help from a wide range of health professionals. During pregnancy, some signs that a baby may have Edwards syndrome may be visible by ultrasound. If these signs are present, it is important that they are investigated further.

How to interpret a high probability result for Trisomy 18

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

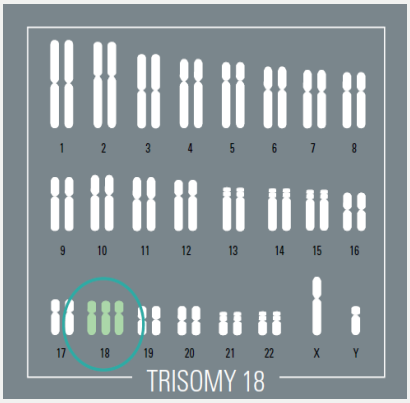

Figure 1 Karyotype showing a person with three copies of chromosome 18. This person would have Edwards syndrome.

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. Trisomy 18;2015 Jan 8
- Centre for Genetics Education (AU). Fact sheet 38 -Trisomy 18 Edwards syndrome [Internet]. 2016 Aug
- National Library of Medicine (US). Genetics Home Reference [Internet]. Bethesda (MD): The Library; 2017 Nov 14. Trisomy 18; [reviewed 2012 Mar]

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

14 Giffnock Avenue, Macquarie Park, NSW 2113, Australia
T 1800 010 447 | **E** info@sonicgenetics.com.au
www.sonicgenetics.com.au

May 2018

SHG-MKT-0040-00.1