Sonic Genetics
Quality is in our DNA

harmony®
PRENATAL TEST

Trisomy 21
Down 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.

Down syndrome is a condition that occurs when there is an extra copy of chromosome 21, meaning there are three copies instead of the expected two (Figure 1). It is a common chromosomal condition and occurs in about one in every 700 babies. While the chance of having a baby with Down syndrome increases as women age, it can occur in any pregnancy. There are many different features associated with Down syndrome, however, not everyone will have the same features or severity of some symptoms. Accordingly, the effect of Down syndrome varies from person to person. During pregnancy, it is not possible to know exactly how Down syndrome will affect each individual.

While intellectual disability is a feature of Down syndrome, early intervention programs have been shown to be effective in supporting people with Down syndrome to reach their full potential and lead productive lives. Medical conditions, such as heart defects, are more common in people with Down syndrome and can affect life expectancy, however, improved access to medical care means that most individuals will live into their 60s.

How to interpret a high probability result for Trisomy 21

Receiving a high probability result for trisomy 21 means that there is an increased chance that your baby will have Down 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. For additional information or to be connected with other families you can contact Down Syndrome Australia at downsyndrome.org.au or 1300 881 935.

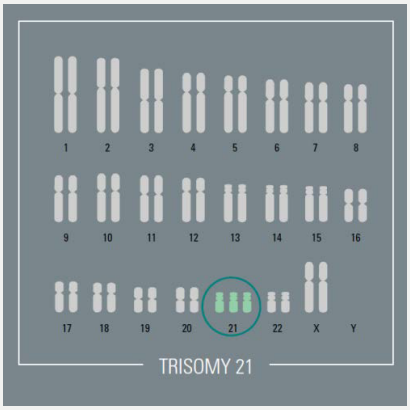

Figure 1 Karyotype showing a person with three copies of chromosome 21. This person would have Down syndrome.

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

- O'Connor, C. Trisomy 21 Causes Down Syndrome. *Nature Education* 2008; 1(1):42
- Down Syndrome Australia. Prenatal testing for Down syndrome [Internet]. 2017 Jun
- Centre for Genetics Education (AU). Fact sheet 36 - Trisomy 21 Down syndrome [Internet]. 2016 Aug
- National Library of Medicine (US). Genetics Home Reference [Internet]. Bethesda (MD): The Library; 2017 Oct 17. Down syndrome; [reviewed 2012 Jun]

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

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