Sonic Genetics
Quality is in our DNA

harmony®
PRENATAL TEST

22q11.2 deletion 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.

22q11.2 deletion is the most common chromosome microdeletion and is often referred to as DiGeorge syndrome. It occurs when there is a very small deletion in a recognised region of chromosome 22 (Figure 1). 22q11.2 deletion syndrome occurs in about one in 1,000 to one in 4,000 pregnancies. Unlike some chromosomal conditions, the chance of having a baby with 22q11.2 deletion syndrome does not increase as women age; it can occur in any pregnancy. In most people with a 22q11.2 deletion, it has occurred by chance and has not been inherited. A small proportion of people with 22q11.2 deletion will have inherited the deletion from a parent with the same chromosome deletion. Testing both parents can help to determine the likelihood of the condition happening in other pregnancies.

A deletion in 22q11.2 region can affect many different parts of the body, including the heart, the immune system and the endocrine system. 22q11.2 deletion syndrome can also affect intellectual ability, speech and can lead to psychiatric illness in young adults. The features of someone with 22q11.2 deletion syndrome are extremely varied; even in families in which a number of people have the same deletion. Prenatal detection of 22q11.2 deletion can allow for early medical and developmental intervention which may improve the outcomes of quality of life of a person with the condition.

How to interpret a high probability result for 22q11.2 deletion syndrome

Receiving a high probability result for 22q11.2 deletion means that there is an increased chance that your baby will have 22q11.2 deletion 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. Rarely, having a high probability result could mean that the condition is present in the mother. This is called an incidental finding. 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 and require specialised tests for looking for smaller deletions known as FISH or microarray testing. 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 22q Foundation Australia & New Zealand at 22q.org.au.

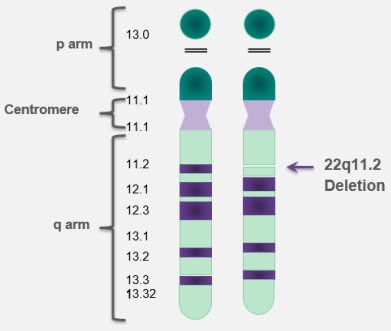

Figure 1 One chromosome 22 showing a small deletion of the 22q11.2 region. This person would have 22q11.2 deletion syndrome.

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

- Rauch et al. *Am J Med Genet* A. 2006 Oct 1;140(19):2063-74
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- Bassett et al. *J Pediatr.* 2011 Aug;159(2):332-9
- McDonald-McGinn et al. Gene Reviews(2013)
- Orphanet Report Series –Prevalence and incidence of rare diseases. 1, Jan 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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