22q11.2 Deletion Syndrome

22q11.2 deletion is the most common chromosome microdeletion. It occurs when there is a very small deletion in a recognised region of chromosome 22 (figure 1). 22q11.2 deletion syndrome is thought to occur in about 1 in 1000 to 1 in 4000 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. While for the majority of people who have a 22q11.2 deletion it happens by chance, a small number of people may inherit the deletion from a parent who also has the condition. 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 could lead to psychiatric illness in young adults. The features of someone with 22q11.2 deletion syndrome are extremely varied, even members of the same family will not always be affected in the same way. Prenatal detection of 22q11.2 deletion syndrome can allow for early medical and developmental intervention which may improve the outcomes of quality of life of a person with the condition.

What does a Non-invasive Prenatal Test (NIPT) result mean?

Receiving a high probability result for 22q11.2 deletion means that there is a significant chance that a baby will have 22q11.2 deletion syndrome. However, NIPT is a screening test meaning in some rare instances, the baby may not actually have the condition. There are many different reasons for these results to be different and some of them cannot be avoided. 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.

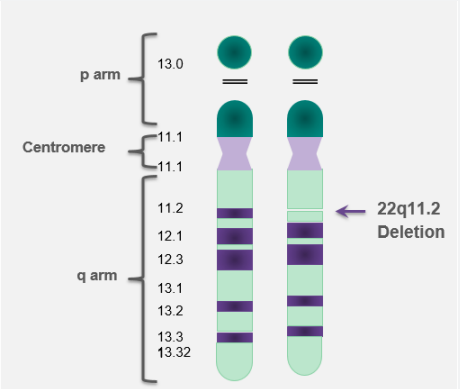

Figure 1 One chromosome 22 showing a small deletion of the 22q11.2 region.This person would have 22q11.2 deletion syndrome

It is important for each individual or family to understand what these results mean for them and their situation. Your healthcare provider should be able to answer a number of questions and for additional information or to be connected with other families you can contact 22q Foundation Australia & New Zealand at 22q.org.au.

This information sheet provides general information about the test result and recommendations. It is not intended to replace genetic counselling by a medical professional.

Non-invasive prenatal testing (NIPT) based on cell-free analysis is not diagnostic: results should be confirmed by diagnostic testing. Before making any treatment decisions, all women should discuss their results with their healthcare provider, who can recommend confirmatory, diagnostic testing where appropriate.

Reference Material:
- Rauch et al. Am J Med Genet A. 2006 Oct 1;140(19):2063-74.
- McDonald-McGinn et al. Genet Couns. 1999;10(1):11-24.
- McDonald-McGinn et al. Nature Reviews Disease Primer. 2015 Nov 19.
- 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.