Trisomy 21 - Down Syndrome

Down syndrome is a well understood 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 1 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 with Down syndrome will have them all and the effects will vary from one person to another. 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 very 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 the life expectancy however, improved access to medical care means that most individuals will live into their 60s.

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

Receiving a high probability result for trisomy 21 means that there is a significant chance that a baby will have Down 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. 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.

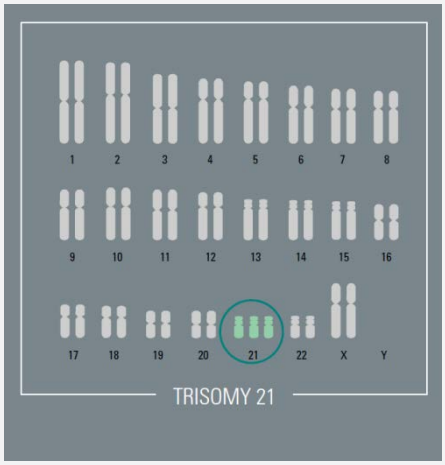

Figure 1 Karyotype showing a person with three copies of chromosome 21. This person would have Down syndrome.

It is important for each individual or family to understand what these results mean for them and their situation. A 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 Down Syndrome Australia at downsyndrome.org.au or 1300 881 935.

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:
- 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]