Trisomy 13 - Patau Syndrome

Patau syndrome, otherwise known as trisomy 13, is a very serious chromosomal condition that occurs when there is an extra copy of chromosome 13, meaning that there are three copies instead of the expected two (figure 1). Patau syndrome occurs in about 1 in 16,000 newborns and unfortunately most babies with Patau syndrome will miscarry and those babies that are born, most will live for just a short time. While the chance of having a baby with Patau syndrome increases as women age, it can occur in any pregnancy.

Patau syndrome can affect many different parts of the body including the heart and kidneys, brain development, and other physical abnormalities. Patau syndrome is also associated with failure to grow and severe intellectual disability. There is no cure for Patau syndrome and the symptoms can be very 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 Patau syndrome may be visible by ultrasound. If these signs are present, it is important that they are investigated further.

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

Receiving a high probability result for Patau syndrome means that there is a significant chance that a baby will have Patau 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.

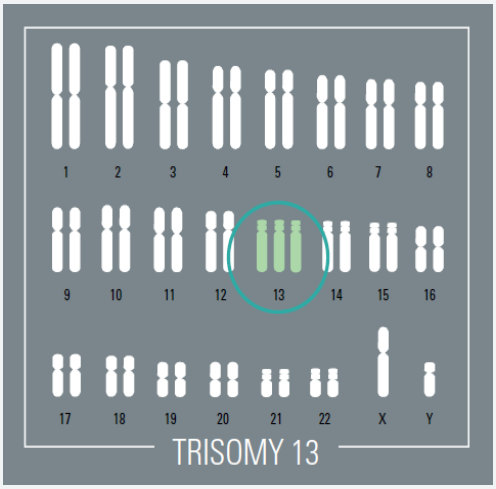

Figure 1 Karyotype showing a person with three copies of chromosome 13. This person would have Patau syndrome.

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 you may have about the condition and your results. Further information can be found by visiting the references below.

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.

References Material:

- A.D.A.M. Medical Encyclopedia. Atlanta (GA): A.D.A.M., Inc.; ©2005. Trisomy 13;2015 Jan 8.
- Centre for Genetics Education (AU). Fact sheet 37 -Trisomy 13 Patau syndrome [internet]. 2016 Aug.
- National Library of Medicine (US). Genetics Home Reference [Internet]. Bethesda (MD): The Library; 2017 Oct 17. Trisomy 13; [reviewed 2013 Nov]