Trisomy 18 - Edwards Syndrome

Edwards syndrome, otherwise known as Trisomy 18, is a very serious chromosomal condition that occurs when there is an extra copy of chromosome 18, meaning that there are three copies instead of the expected two (figure 1). Edwards syndrome occurs in about 1 in 5000 newborns and unfortunately most babies with Edwards syndrome will miscarry and those babies that are born with Edwards syndrome, most will live for just a short time. While the chance of having a baby with Edwards syndrome increases as women age, it can occur in any pregnancy.

Edwards syndrome can affect many different parts of the body including abnormalities of the heart, brain, kidneys and gastro-intestinal system. Edwards syndrome can also cause failure to grow, development delay, intellectual disability and other physical features. There is no cure for Edwards 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 Edwards 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 Edwards syndrome means that there is a significant chance that a baby will have Edwards 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.

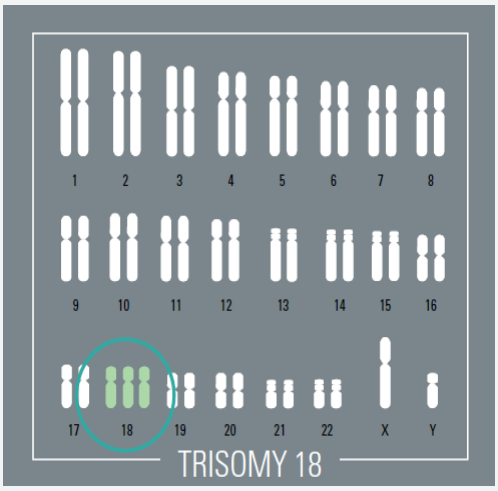

Figure 1 Karyotype showing a person with three copies of chromosome 18. This person would have Edwards syndrome.

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.

Reference Material:

- A.D.A.M. Medical Encyclopedia. Atlanta (GA): A.D.A.M., Inc.; ©2005. Trisomy 18;2015 Jan 8.
- Centre for Genetics Education (AU). Fact sheet 38 -Trisomy 18 Edwards syndrome [internet]. 2016 Aug.
- National Library of Medicine (US). Genetics Home Reference [Internet]. Bethesda (MD): The Library; 2017 Nov 14. Trisomy 18; [reviewed 2012 Mar]