Information leaflet for the parents of a patient

Genomic testing

The doctor has suggested that your child should have genomic testing. In this leaflet you will find information about this test.

Why this test?
Your child's condition may be caused by a variant in the DNA. The aim of the DNA test is to trace this DNA variant.

What is needed for the test?
For this test a blood sample will be taken from your child. Your child can eat and drink normally before the blood test. The DNA will be taken from the blood sample. A blood sample from the biological parents is usually also necessary in order to compare the DNA between parents and child.

Types of test

- **Gene panel:** a DNA test that examines a certain number of genes that are known to be linked to your child's condition.
- **WES** (whole exome sequencing): a DNA test that examines all genes.
- **WGS** (whole genome sequencing): a DNA test that examines all genes and the DNA in between the genes.

Potential outcomes

1. **The cause is found**
   A DNA variant is found that is the cause (or most likely the cause) of your child's condition. This gives more information about heritability. It is sometimes possible to predict how your child's condition will develop in the future and if treatment is possible.
2. **No cause is found**
   No DNA variant is found that could be the cause of your child's condition. There could be two reasons for this:
   1. There is no DNA variant that is the cause of your child's condition.
   2. The DNA variant causing your child's condition cannot be proved yet with the current knowledge of genetics. Your doctor will talk with you about the possibility of further testing.
3. **The result is unclear**
   A variant of unknown significance is found, what this means is unclear. It may mean nothing. It is sometimes helpful to test other members of the family. It is up to you to decide if you wish to ask members of your family to cooperate with this.
4. **Incidental findings**
   Sometimes a DNA variant is found that is not the cause of your child's condition, but that does play a role in another genetic condition. This is called an incidental finding. The chance of finding an incidental finding is small. This chance is the smallest in the gene panel test. Your doctor will give you a number of examples of incidental findings. If there are certain incidental findings that you either do or do not wish to have information about, discuss this with your doctor.

What this test cannot do
This test will not find all genetic conditions. The DNA test searches for the cause of your child's condition. Sometimes, a variant related to another condition (incidental finding) that has not been looked for is found.

Family connections
If the DNA of the parents is also examined, it can be seen if the parents are the biological parents of the child.

Consequences for the family
The test result may sometimes also be important for your family, now or in the future. It is possible that they could also have the condition. Or their children (or future children) could also get the condition.

When and how will I get the results?
Your doctor will discuss with you when and how you will get the results.

Reimbursement
The costs will be reimbursed by your health insurance. You can ask your own insurance company about this. You only pay the deductible if this has not already been used up for that year. Children under the age of 18 years do not have a deductible. If you also have had a DNA test to compare your child's DNA with yours, then this is covered by your child's health insurance.

Will the DNA test have consequences for my insurances?
There are other insurances besides health insurance. Disability insurance and life insurance, for example. More information on insurances and heritability can be found on www.erfelijkheid.nl. You can ask your own insurance company or your advisor about this.

After the results

- Contact with fellow patients
  You can find people with the same condition through patient organisations (see www.zichtopzeldzaam.nl) and Facebook groups. Sometimes your doctor will be able to arrange contact with fellow patients.
    - Platform ZON (www.ziekteonbekend.nl) for parents of chronically ill children without a diagnosis or with a very rare diagnosis.
    - ZeldSamen (www.zeldsamen.nl), networks for rare genetic syndromes.
    - (Sch)ouders (www.schouders.nl) is a platform for and by parents of a child with a physical and/or intellectual disability, chronic illness and/or a developmental disorder.
- Counselling
  A counsellor is available to help you to process the results. Your doctor will be able to refer you.
- More information about DNA testing
  See www.erfelijkheid.nl
- Testing in the future
  The possibilities of DNA testing will increase in the future. Sometimes, the cause of the condition will be found a few years in the future, or more may become known about the DNA variant.
    - Generally, the DNA is not tested again. If you wish, in a few years you could get into contact again to see if any new possibilities have become available in the meantime.

- If new possibilities or results do become available, sometimes the doctor may contact you again. If you do not want this, please let us know (see consent form).

What happens to your DNA?

- The DNA is stored at the Genetics department. It is stored in accordance with legal requirements. For example, the data will not be shared with insurers.
- By sharing information about DNA with other doctors or researchers, more information about the consequences of a DNA variant will become available. This is especially important in rare conditions. For this reason, DNA variants are shared with other national and international laboratories. Names are not used, so no-one can see who the DNA belongs to.
- If you wish, together with your child, you can give permission for the DNA to be used for further scientific research (see consent form).

Questions?

If you still have questions after reading this information leaflet, or if you want to change your consent, please contact your doctor on the Clinical Genetics Department of the [name of hospital] via [tel: email address]. For further information about the department see [hospital website].