Patiëntinformatie

Maastricht UMC+

Genomic diagnostic testing (information for parents of a patient)

Your doctor has suggested that your child undergoes genomic diagnostic testing. More information about this testing is provided below.

Why this test?

Your child's disorder/condition may be caused by a pathogenic variant in the DNA (a genetic alteration that directly contributes to the development of the disorder). The purpose of the genomic diagnostic testing is to trace this pathogenic variant.

What is required for testing?

Your child will be required to provide a blood sample but does not need to follow a nil-by-mouth regime for this. DNA will be extracted from this blood sample. You as a parent might also be asked to provide a blood sample to compare your DNA with that of your child.

Which test?

- **Gene set**
  A genomic diagnostic test for several genes known to be a cause of your child's disorder.
- **WES (whole exome sequencing)**
  A genomic diagnostic test in which all genes are examined.
- **WGS (whole genome sequencing)**
  A genomic diagnostic test in which all genes and the DNA material in between the genes is examined.

What are the possible outcomes?

Cause found
A pathogenic variant is found in the DNA that is probably the cause of your child's disorder. This tells us more about the heredity aspect. Sometimes a prediction can be made on how your child's disorder will develop in the future and whether treatment is possible.

No cause found
No pathogenic variant was found in the DNA that could be the cause of your child's disorder. There are two possible reasons for this:

1. There is no pathogenic variant in the DNA that causes the disorder.
2. There is a pathogenic variant in the DNA, but it cannot yet be identified with the current test. The doctor will discuss with you about whether there are any options for further testing.

Inconclusive result
A variant in the DNA is found, but it is not clear if this is the cause of your child's disorder. It could be something of no significance, and it might be helpful to test other family members. There is no obligation, and you are completely free in asking your family members for their cooperation.

Incidental findings

In addition to the above results, a pathogenic variant could be found in the DNA, which is not the cause of your child's disorder, but does play a role in another hereditary disease. This is called an incidental finding. There is only a slight chance of identifying any incidental findings and your doctor will provide you with some clear examples.

What types of incidental findings are there, and which ones will be reported to you?

1. The predisposition to a disorder **will** be reported to you if medical treatment or monitoring for your child is possible.
   You can opt out if you do not wish to be informed of this.
   It is not possible to opt out for children under 12 years of age with a clinical feature that manifests itself in childhood.
2. The predisposition to a disorder is **not** reported to you if (based on the current data and information on the condition in question) **no** medical treatment or monitoring is possible. You can, however, opt in if you wish to be informed of this.
   It is not possible to opt in for children under the age of 16.
3. If there is a high risk of a disorder in any unborn children of yours or your child's (25% or higher), you **will** be informed of this. You can opt out if you do not wish to be informed of this.
   Parents must be unanimous in their choice of whether to opt out.

What are the limitations of this test?

This test is unable to identify all hereditary disorders. The DNA test looks for the cause of your child's disorder. There are occasions when something else is found - an incidental finding - but this is not something that is actively sought for.

Implications for family members

The results may also be of significance for your family members, now or in the future. Perhaps they or their (unborn) children are at an increased risk of inheriting the disorder. If this is the case, your doctor will provide you with some information to be shared with your family.

When can I expect the results?

The doctor will inform you on how and when you will receive the results.

Insurance cover

The costs of the tests are covered by your health insurance, (with the exception of some budgetary stipulations). You can ask your own health insurance provider for more details. Children under the age of 18 do not pay any excess. In the event that a DNA test is also being performed on you to compare your child 's DNA with yours? This is covered by your child's health insurance.

Will the DNA test have any consequences for my insurance policies?

Heredity testing can sometimes affect the ability to take out some insurance policies, such as disability insurance or life insurance. More information about insurance and heredity can be found at www.erfelijkheid.nl. You can also ask your insurer or insurance advisor.

Future contact

On completion of a genomic diagnostic test, there is no further active search for the cause of the disorder. However, it is possible that new data or information, important to you, will become available in the future.

- If no cause has been found, you can contact our department again after 3-5 years. A decision can then be made on whether it may be useful to conduct fresh analysis on the data from the genomic diagnostic test.
- In rare cases, the department of Clinical Genetics will contact you itself. If you wish to opt out from this, you can do so in the consent form.

The costs of any follow-up tests are declared to the health insurer.

What happens to the DNA?

- If necessary, your child's DNA will be sent to another national or international laboratory to perform the genomic diagnostic test. The privacy of your child's personal data is guaranteed during this process.
- The DNA is stored at the Genetics Department in accordance with the statutory regulations. The data will not be passed onto insurers, for example.
- To interpret the test results as accurately as possible, they may be shared with other national or international laboratories. The results are encrypted, which means that the name and date of birth are replaced by a code. Your child's details can only be traced back to the laboratory that carried out the test. This guarantees the privacy of personal data during the process.
- You (together with your child) can give permission for the DNA to be used for further scientific research. Your child’s data will be handled with due diligence. You do not directly benefit from this. On rare occasions a researcher may discover something that could be important to your health or that of your family members. The doctor will inform you about this.

Contact

If you have any questions after reading this information or would like to change your consent status, please contact the doctor at the Clinical Genetics Department.

Maastricht UMC+
Outpatient clinic Clinical Genetics
T: (+31) (0)43.3875855
E: polikliniek.klinischegenetica@mumc.nl
Website: klinischegenetica.mumc.nl

Laatst bijgewerkt op 24 februari 2022