Genetic testing
Information for parents of patients

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

Why this test?

Your child's disorder/condition might be caused by a change in the DNA. The purpose of the genetic testing is to identify this change. Identification of the change in the DNA that causes the condition might help with treatment, monitoring and family planning decisions.

What is required for testing?

Your child will be required to provide a blood sample and DNA will be extracted from this 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 diagnostic test for several genes. Changes in these genes are known to cause your disorder.
- **WES** (whole exome sequencing): A diagnostic test in which all genes in the genome are examined.
- **WGS** (whole genome sequencing): A diagnostic test in which the complete genome is examined (all the genes and all the DNA material in between the genes).

What are the possible outcomes?

1. Cause found: A change in the DNA was identified that is (likely to be) the cause of the disorder. This can tell us more about the inheritance pattern of the disorder in the family. Sometimes a prediction can be made about how the disorder will develop in the future, what monitoring is required and whether treatment is possible.
2. No cause found: No change in your DNA was identified that is (likely to be) the cause of the disorder. There are two possible reasons for this:
    1. The disorder is not caused by a change in the DNA.
    2. The disorder is caused by a change in the DNA that cannot be identified with the current test. The doctor will discuss whether there are any options for further testing.
3. Inconclusive result: A change in the DNA was identified, but it is not clear if this is the cause of disorder. It could be something of no significance and it might be helpful to test other family members and/or perform additional tests. There is no obligation, and you are completely free to ask your family members for their cooperation.

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Unsolicited findings

In addition to the above results, it is possible that a change in the DNA may be identified that is not the cause of the disorder, but instead plays a role in another genetic disorder/condition. This is called an unsolicited finding.

What types of unsolicited findings are there, and which ones will be reported?

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 knowledge of 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 the test?

The test is used to try and identify the change in the DNA that causes the disorder. However, the test is unable to identify all of the possible changes in the DNA and it is possible that the test will not identify the cause of the disorder.

Implications for family members

The results may also be of significance for other members of your family, now or in the future. They, or their (unborn) children, might have an increased risk of inheriting the disorder. If this is the case, your doctor will provide you with information that you can share with your family.

When can I expect the results?

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

Insurance cover

In most cases, the costs of the test should be covered by the health insurance. Ask your health insurance provider for details. You only pay your own risk if it has not yet already been used-up for that year. In the event that parent's DNA will also be tested to compare the DNA, parents will not have to pay themselves for these costs.

Will the DNA test have consequences for insurance policies?

Genetic testing can affect the ability to take out some types of insurance policy, such as disability insurance or life insurance. More information please contact your insurer or insurance advisor.

Future contact

On completion of the genetic test, no further active search for the genetic cause of the disorder will be carried out. However, it is possible that in the future, new data or information which are important, will become available.

- If no cause for the disorder was identified, you can contact our department again after 3-5 years. A decision can then be made on whether it would be useful to perform a new analysis of the genetic data from the test.
- In some cases, the Department of Clinical Genetics will contact you with new information. If you wish to opt out from this, you can indicate this on the consent form.

The costs of any follow-up tests will be declared to the health insurer and may be charged. Please check with your insurance provider.

What happens to the DNA?

- If necessary, the DNA will be sent to another (inter)national diagnostic laboratory to perform the test. The privacy of personal data is guaranteed during this process.
- The DNA and the data will be stored at the Department of Clinical Genetics, in accordance with the statutory regulations. DNA or the data will be shared with other parties, after receiving consent to do so.
- To interpret the test results as accurately as possible, the data might be shared with other (inter)national laboratories. In such cases, the results will be encrypted, which means that the name and date of birth are replaced by a code. Personal details can only be traced back to the laboratory that carried out the test. This guarantees the privacy of personal data during this process.
- You can give permission for the DNA to be used for scientific research. Data will be handled with due diligence. Although it is unlikely to benefit directly from this, on rare occasions a researcher might discover something that could be important for the health of your child or that of other family members. The doctor will inform you about this.

Questions?

If you have any questions after reading this information or would like to change your consent status, please contact your consultant clinical geneticist at the Department of Clinical Genetics at the Erasmus Medical Center: 010 703 6915 or ervo@erasmusmc.nl. For more information about the department see www.erasmusmc.nl.