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Information Sheet on the Haemoglobinopathy Test

What are haemoglobinopathies?

Haemoglobinopathies are diseases caused by disorders of the red blood pigment (haemoglobin) in red blood cells. Haemoglobinopathies are usually inherited diseases and result in disease patterns of variable severity depending on the specific genetic defect involved.

The World Health Organization (WHO) estimates that about seven per cent of the world's population carry a gene mutation that manifests clinically as a haemoglobinopathy. According to WHO estimates, around 300,000 to 500,000 children are born with a severe form of haemoglobinopathy every year.

Several different diseases make up the group of haemoglobinopathies, the most significant of these being the **thalassaemias** and **sickle cell disease**. These are inherited diseases.

Why might a haemoglobinopathy test be necessary in connection with cord blood donation?

Blood stem cell transplants are sometimes used to treat severe forms of these diseases, so it is important that the frozen cord blood units be tested for these diseases before their use. This can be done with a haemoglobinopathy test, which is a genetic test that indicates whether someone has a a genetic disposition for these diseases.

These genetic tests need not be performed on all frozen umbilical cord units, but only on those that match a particular patient according to the tissue typing results. Thus, they are performed only at the time when the frozen umbilical cord unit is requested for transplantation. Haemoglobinopathy testing can yield results that are of significance for your child and your child's descendants. There is no need to take an additional blood sample for this test. It can be performed using a sample stored for this purpose at the time of donation. The test would not involve any costs for you.

The samples are preserved and examined in accordance with current scientific and technological standards.

What does this haemoglobinopathy test mean for my child and for me?

We distinguish between two forms of thalassemia and sickle cell disease:

1. **Affected persons** (homozygous^{+}) usually produce only abnormal haemoglobin. This results in severe medical conditions. Affected persons need regular and lifelong medical assistance.
2. **Carriers** (heterozygous*) have both normal and abnormal haemoglobin. Under normal circumstances, this will not affect the structure of their red blood cells and the disease will not become manifest. However, carriers can pass on the defect to their children. When a carrier has a child with someone who is also a carrier, there is a chance that the child will inherit a severe form of the disease.

What happens if the results of the haemoglobinopathy test are abnormal?

1. If your child is **not** capable of judgement at the time of the test (we will assume this to be the case if your child has not yet reached the age of 14)

and is an affected person:

Due to the significance that this result has for the health of your child and of your child's descendants, the law requires us to notify you of the result and requires you to acknowledge it. You will be offered a genetic counselling from a medical specialist after receiving the results. This genetic counselling would not involve any costs for you.

and is a carrier:

Although this result is not of direct significance for the health of your child, it may be of significance for your child's descendants. When the test is requested, you will be asked whether you wish to be notified if the test reveals that your child is a carrier. You will have the option of genetic counselling from a medical specialist at the time of the test as well as after receiving the results. This genetic counselling would not involve any costs for you.

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2. If your child is **capable** of judgement at this time (we will assume this to be the case if your child is 14 years old or older)

Your child will receive the required information about haemoglobinopathies at the time of the test. Directly thereafter, your child will decide whether he or she wishes to be notified of the test results. Children capable of judgement are entitled to decide for themselves as to whether they wish to know the results. They have a right "not to know", even if the result is of significance for their own health. Your child will have the option of genetic counselling from a medical specialist at the time of the test as well as after receiving the results. This genetic counselling would not involve any costs for you and/or your child.

Unfortunately, a cord blood donation will not be possible if you do **not** consent to a genetic test of this kind.

[+]homozygous: homozygous individuals have the gene defect in both genes.

*heterozygous: heterozygous individuals have the gene defect in only one gene.