Bijlage 4: Patiëntenvoorlichting Engels

RTA Hemoglobinopathiescreening preconceptioneel en tijdens zwangerschap

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Carrier information for hemoglobinopathies
If you are reading this leaflet, your obstetric caregiver has made you aware of the possibility of carrier testing for hemoglobinopathies.
In this leaflet we will give you more information about this.

Hemoglobinopathy
Hemoglobinopathies are hereditary defects of the hemoglobin. They are also known as 'hereditary anemia'. The most common forms of hemoglobinopathies are sickle cell disease and thalassemia. Hemoglobine is a proteine contained in red blood cells. Red blood cells are part of the blood. Hemoglobin picks up oxygen from the air in the lungs. After that, hemoglobin delivers the oxygen to the organs and other parts of the body.

In someone with hereditary anemia, the hemoglobin is not produced properly. This means that the shape of the hemoglobin is slightly different (as is the case in sickle cell disease) or the composition of hemoglobin has changed (as seen in thalassemia). This makes the red blood cells look slightly different or too little hemoglobin is produced. The body recognizes the abnormal red blood cells and breaks them down faster than regular red blood cells. This causes a person to develop chronic anemia, causing less oxygen to be transported from the lungs to the rest of the body.

Anemia causes feelings of fatigue, headache and shortness of breath. Depending on the cause of anemia, there can also be serious consequences such as jaundice, damage to organs and growth retardation.

Increased risk of hemoglobinopathies
Hemoglobinopathies are hereditary diseases. A child has a chance of developing a hemoglobinopathy (or being a carrier of a hemoglobinopathy) if one or both parents are carriers of the disease. Being a carrier generally means that parents themselves do not know that they carry the disease because they have no or only mild symptoms.
However, they can pass on the disease to their children.
The risk of being a carrier is greatly increased if a hemoglobinopathy (or being a carrier) runs in the family, but also if someone originally comes from Africa, the Antilles, the Mediterranean region, Southeast Asia or the Middle East. In these areas the carrier frequency varies between 1 in 15 people to 1 in 7 people. In some African countries, even 30-40% of the population is a carrier.

Testing for hemoglobinopathy
It is possible to test for the carrier status of a hemoglobinopathy in (future) parents. This test is initially offered to the parent with an increased risk of being a carrier for a hemoglobinopathy. If this parent is found to be a carrier, the other parent will also be examined in order to determine the risk of disease in the child. If both parents have an increased risk of a hemoglobinopathy, then both parents may be tested at the same time.
The results of the tests are expected within four weeks.

If both parents are found to be carriers of a hemoglobinopathy, a meeting will take place with a clinical geneticist in the hospital to discuss the risks and possible follow-up examinations for the unborn child.

Cost
Testing for hemoglobinopathy is requested by a midwife, general practitioner or gynaecologist. The laboratory cost for this test is € 100 to € 300. The cost for the tests is covered by health insurance. However, the deductible is used for this.