

Title: Functional and genetic characterization of Danish patients suspected of hereditary anemia (DAHEAN)

We will ask if your child would like to participate in a scientific study.

Participation in the study is voluntary. You can withdraw your consent at any time and without giving a reason.

The study is a national collaboration coordinated by Rigshospitalet, Blegdamsvej 9, DK-2100 Copenhagen.

The study is led by:

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With the study, we would like to

1. Make sure your child receives the correct diagnosis of their deficiency of red blood cells (often called anemia)
2. Make sure that we know your child's illness as best as possible and thereby offer the best possible treatment

Plan for the study

- We collect the child's health information in a secure electronic database.
- In collaboration with the child's hospital doctor, a group of Danish anemia experts will assess the health information..
- It may be that the experts suggest further examinations to give the child a better diagnosis.
- It may also be that the experts have suggestions for treating the child's illness.

The child does not need to have any additional tests to participate in the study. However, it may be that the experts in anemia have suggestions for extra tests (e.g. blood tests) that can help us to know the child's disease in the best possible way. The child's own doctor will discuss any extra examinations with parents/guardians and, as far as possible, also the child.

On the following pages, you can see in more detail what the study is about and what it means if the child participates in the study.

1 PARTICIPANT INFORMATION ABOUT PARTICIPATION IN A SCIENTIFIC STUDY

Title of the study: Functional and genetic characterization of Danish patients suspected of hereditary anemia (DAHEAN)

We would like to ask if your child would like to participate in a national scientific study coordinated by Dr. Andreas Glenthøj at Rigshospitalet.

Before you decide whether your child should participate in the study, you must fully understand what the study is about and why we are conducting the study.

You will be invited to a conversation about the study, where you can ask the questions you have about the study. You are welcome to bring a family member, friend or acquaintance to the meeting.

If you decide that the child participates in the study, we will ask you to sign a declaration of consent, which is a paper where you agree to the child participating in the study.

You have the right to a period of reflection before deciding whether the child will participate.

Participation in the study is completely voluntary. You can choose to withdraw your child from the study at any time and without giving a reason. There will be no consequences for the child's further treatment.

2 WHY DO WE RESEARCH ANEMIA?

Many people in the world are born with a disease that causes anemia. This is due to a change in one of the genes that the body needs to use to make red blood cells. Some patients have very mild anemia and do not notice it in daily life. Other patients may be more tired and have difficulty playing sports. Some even need to be treated with blood transfusions.

It is often difficult to find out exactly why a patient has anemia. This makes it difficult to choose the right treatment.

With this study, we would like to

- 1) Ensure that the child receives the correct diagnosis of his or her anemia
- 2) Make sure that we know the child's illness as well as possible and thereby can offer the best possible treatment

3 SAMPLES FROM THE CHILD - RESEARCH BIOBANK

If the child participates in the study, we register the examinations that the child has undergone. For example, it can be the result of blood tests. The study does not require the child to have extra tests done and it is not dangerous to participate.

If there is material in surplus from a sample (e.g. excess blood), it can be stored until the study is completed in 2037. This makes it easier to carry out extra tests without us having to take extra samples from your child.

If there is material in surplus after the end of the study in 2037, it will be stored for possible use in future research projects. However, this requires specific approvals. We must ask you, or your child if they have

reached the age of majority, and obtain approval from the Research Ethics Committee. In rare cases, the Research Ethics Committee can give permission for us not to ask the patient for permission. You can contact us at any time to have material that we have stored from the child destroyed.

Rarely are there very special tests that we cannot do in Denmark. Here we will be assisted by a laboratory in the Netherlands. If we send material (e.g. blood) abroad, it will be destroyed after it has been tested.

4 PLAN FOR THE STUDY

If the child participates in the study:

- 1) We will store information about the child and the examinations that have been carried out on the child in a secure database
- 2) It may be that the child's own hospital doctor offers to carry out extensive examinations of the child's genes. Then the child's doctor will ask you for permission to do so, as extensive testing of genes requires consent.
- 3) We may recommend additional tests, for example blood tests, to help give the child a better and more certain diagnosis of their anemia
- 4) We will discuss health information (e.g. blood test results) with a number of experts in anemia to give the child the best and most reliable diagnosis

Through genetic testing (DNA), we can often get a precise explanation as to why the child has anemia. This can help us plan the best possible treatment for the child's illness.

We only look at genes that can cause poor formation of red blood cells. However, there is still a small risk that we will find genetic defects that are important for something other than the formation of red blood cells. If this happens, we will offer you a conversation with an expert about this (genetic counselling).

The genetic studies are done by the National Genome Center and you can read more about it on their website (www.ngc.dk).

Data from the child's genetic tests are stored securely and are protected by legislation (the General Data Protection Regulation and the Data Protection Act).

5 USEFULNESS OF THE STUDY

The study tries to improve our ability to diagnose congenital anemia. In this way, the individual patient gets:

- 1) A better and more accurate diagnosis
- 2) The best basis for choosing the right treatment

In the future, we expect that there will be more and more targeted treatments for congenital anemia, which will make it even more important to know the right diagnosis.

By becoming better at diagnosing your child's anemia, we also hope to become better at helping other patients with the right diagnosis and the right treatment in the future.

6 SIDE EFFECTS, RISKS, COMPLICATIONS AND DRAWBACKS

It is not dangerous to participate in the study. We only register the examinations that you arrange with the child's own hospital doctor. Typically, we register the results of blood tests and any bone marrow examinations.

The child does not need to have more examinations just because he or she participates in the study. It may be that the child's own doctor becomes aware of more examinations that can help the child get the right diagnosis.

As far as possible, the child's own doctor tries to take extra blood samples at the same time as blood samples are taken anyway. You can get a bruise after blood sampling. It disappears after a few days. Others may feel anxious of having blood samples taken, and here the staff at the child's ward will help you in the best possible way.

Bone marrow examination is not performed due to participation in the study. Children who have a bone marrow examination are anesthetized and sleep while it is done.

	Frequent/not severe	Rare/serious	Long-lasting
Side effects	None	None	None
Risks	None	None	None
Complications	Bruising at the site where the needle is inserted.	None	None
Disadvantages	Discomfort/pain when taking blood samples	None	None

If you/you/the child experience problems during the study, you are very welcome to tell us or the child's doctor.

7 STANDARD OF CARE OUTSIDE THE STUDY

If you do not participate in the study or that you choose to withdraw from the study, it is perfectly fine. The child still has the right to receive completely normal treatment – both now and in the future.

8 INFORMATION FROM THE CHILD'S MEDICAL RECORD

If the child participates in the study, the study coordinator and his/her colleagues will have the opportunity to look at the medical records. Selected information from this is entered into a secure database in the Capital Region of Denmark that has been approved for the purpose (by the Danish Data Protection Agency).

We use the medical record information to:

- 1) Be able to present and discuss the child's health information with experts in anemia
- 2) Be able to ensure the best and most accurate diagnosis of the child's anemia

We will look for the following information:

- Identification information: Name, birthday, CPR number, contact information
- Genetic data: The result of genetic analyses relevant to anemia
- Health information: Answers to tests (e.g. blood tests), symptoms, treatment
- Family origin/ancestry may be relevant because some hereditary anemias are more common in certain populations.
- Whether there is stored biological material (e.g. excess blood) that can be used if more tests are needed

It is also a requirement that information from the child's medical records must be able to be reviewed by persons who are to carry out statutory quality control of the study.

9 HOW DO WE TAKE CARE OF THE CHILD'S INFORMATION?

We take good care of the child's information and comply with the legislation in the area (the Data Protection Act and the General Data Protection Regulation).

10 WHO CAN PARTICIPATE?

All patients suspected of having anemia are welcome to participate in the study. If the study does not prove to help patients, we will stop the study.

11 HOW IS THE STUDY PAID?

Consultant Andreas Glenthøj has started the study and is responsible for it.

Andreas Glenthøj is employed at Rigshospitalet but has also done work for and has research collaborations with private companies. When doctors have financial cooperation with private companies, permission must be granted by the Danish Medicines Agency. And these collaborations will always be visible on www.laegemiddelstyrelsen.dk. Some of the companies that Andreas Glenthøj works with are interested in anemia and therefore also interested in the study.

Right now, the study does not receive any financial support. If money is given for the study, it will be reported to the Research Ethics Committee. And you'll always be able to see who supports the study at www.anemia.dk.

Patients do not receive money for participating in the study.

12 HOW DO WE TELL ABOUT THE STUDY?

Whatever we find in the study, we will share the results. We do this by writing articles in international journals and presenting it at meetings in Denmark and abroad.

We will also produce reports showing that the study helps more people get a correct diagnosis and perhaps even better treatment. In the report, we report how many patients have been examined and which

diagnoses we have helped to find. In the report, we only write general information about patients and therefore *not* detailed information about the individual patient. The report will be shared with health authorities and others with an interest in anemia. It can be people, companies or foundations that want to support the study and can thereby see benefit in supporting its expansion.

13 NEED MORE INFORMATION?

We hope that with this information you have received enough information about what the study is all about. And that you know if the child is going to participate. Otherwise, you are very welcome to talk to the child's hospital doctor about this or contact the study coordinator, Andreas Glenthøj.

Best regards



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S13 – Genomics

Consent of the holder of parental responsibility for their child's participation in a health research project that includes extensive mapping of the genetic material

Title of the research project:

Functional and genetic characterization of Danish patients suspected of hereditary anemia (DAHEAN)

Declaration from the holder of parental responsibility:

I/we have received written and oral information and I/we know enough about the purpose, method, advantages and disadvantages of giving my/our consent.

I/we know that participation is voluntary and that I/we can always withdraw my/our consent without my/our daughter/son losing his or her current or future rights to treatment.

I/we are informed that this is a research project that includes extensive mapping of the genetic material. I/we are also informed that in rare cases, clinically relevant changes in my child's genes may be discovered that can lead to a serious illness that can be prevented or treated. I/we can be contacted if necessary.

I/we give consent for _____ (the child's name) participating in the research project and for his / her biological material to be taken out for storage in a research biobank. I/we have received a copy of this consent sheet as well as a copy of the written information about the project for my own use.

The name or names of the holder (s) of custody:

Date: _____ Signature: _____

Date: _____ Signature: _____

Do you want to be informed about the results of the research project itself when the results from all participants are available:

Yes _____ (set x) No _____ (set x)

Statement by the person providing information:

I declare that the subject has received oral and written information about the study.

In my opinion, sufficient information has been provided to enable a decision to be taken on participation in the study.

Name of the person providing information:

Date: _____ Signature: _____