Information for 15-17-year-olds about the study: Functional and genetic characterization of Danish patients suspected of hereditary anemia (DAHEAN).

We would like to ask if you want to take part in a research study.

Taking part is voluntary. You can change your mind and withdraw at any time, without giving a reason. This will not affect your current or future treatment.

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

The study is led by:
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Document version: 15/01/2026 version 2

The purpose of the study is to:

1. Ensure that you get the correct diagnosis of blood disease (anemia)
2. To understand your condition as well as possible, so we can select the best possible treatment

Plan for the study

- We collect relevant information from your medical record and tests in a secure electronic database.
- Together with your hospital doctor, a group of Danish anemia experts will review the information.
- The experts may suggest additional tests if they could help reach a more certain diagnosis.
- The experts may also suggest how to treat your disease best.

You do not need to have any additional tests to participate in the study. However, experts in your disease may have suggestions for extra tests (e.g. blood tests) that can help us to know your disease as best as possible. Your hospital doctor will discuss any additional examinations with you and your parent(s)/guardian(s).

On the following pages, you can see in more detail what the study is about and what it means if you and your parent(s)/guardian(s) agree to participate.

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 you would like to participate in a national scientific study led by Dr. Andreas Glenthøj.

Before you decide whether to participate in the study, you must fully understand what the study is 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 interview.

If you decide to participate, you will be asked to sign a consent form, which is a document confirming your agreement to take part in the study.

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

Participation in the study is completely voluntary. You can choose to stop the study at any time and without giving a reason. There will be no consequences for your 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 changes in the genes that the body uses to produce 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 you get the correct diagnosis of your anemia
2) Make sure that we know your illness as well as possible and thereby offer the best possible treatment

3 SAMPLES FROM YOU - RESEARCH BIOBANK

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

If there is leftover material from a sample (e.g. excess blood), it may be stored until the study is completed in 2037. This allows us to perform additional tests without needing to collect more samples from you.
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 special permission and must ask you and the Research Ethics Committee for permission. 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 any material that we have stored from you 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 you are participating in the study:

1) We will store information about you and the tests/examinations that have been carried out in a secured database
2) It may be that your own hospital doctor offers to do extensive examinations of your genes (genetic material). Then your doctor will ask you and your parents for permission to do so.
3) It may be that we recommend extra blood tests for examinations that can give you a better and more reliable diagnosis of your anemia
4) We will discuss health information (e.g. blood test results) with a range of anemia experts to provide you with the best and most reliable diagnosis

Through genetic testing (DNA), we can often get a precise explanation of why you have anemia. This can help us plan the best possible treatment for your disease.

We will focus on genes related to red blood cell formation. However, there is still a small risk that we may identify genetic changes unrelated to red blood cell formation. If this happens, we will offer you and your parents 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 your 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 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 just register the examinations that you arrange with your own hospital doctor. Typically, we register the results of blood tests and any bone marrow examinations.

You do not need extra tests done just because you participate in the study. It may be that your own doctor becomes aware of several examinations that can help you get the right diagnosis.

As far as possible, your own doctor will try to take extra blood tests at the same time as you have other blood tests. You can get a bruise after blood sampling. It disappears after a few days. Others may be afraid of having blood samples taken, and here the staff at your department 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 experience problems during the study, please let us or your doctor know.

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. You still have the right to receive completely normal treatment – both now and in the future.

8 INFORMATION FROM YOUR MEDICAL RECORD

If you participate in the study, the study coordinator and his colleagues will have the opportunity to look at your 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 information from your medical record to:

1) Be able to present and discuss your health information with experts in anemia
2) Be able to ensure the best and most accurate diagnosis of your 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
- Information about your family's origins, as some types of anemia are more common in specific 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 your medical record 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 your information?

We take good care of your 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 always 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 will share the results by publishing articles in international journals and presenting them 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 write 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 this information helps you understand the study and decide whether you wish to participate. If you have further questions, please speak to your hospital doctor or contact the study coordinator, Dr. Andreas Glenthøj.

Best regards,

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