My name is Dr Timothy Barrett, and I work with your doctor. I am a children's doctor based in Birmingham, UK, and look after people with Wolfram, Alström, Bardet Biedl, and other rare diseases. EURO-WABB is a research study (www.euro-wabb.org). The study will involve young people and adults from across Europe. We will develop a registry (list) of patients with rare diseases like Wolfram, Alström and Bardet Biedl syndromes. By collecting information about these diseases, we hope to learn more about the diseases and what causes them.

An invitation to take part

• We are asking if you would like to take part in the EURO-WABB research study. We are also asking your parent or guardian to agree for you to take part in this study.
• You don’t have to take part unless you want to.
• Before you decide if you want to take part, it is important that you understand why the research is being done and what it will involve for you. Please read the following information carefully and discuss it with your family, friends, doctor or nurse if you want to. Please ask us if there is anything else you would like to know.
• Part 1 tells you what the study is about and what will happen to you if you take part.
• Part 2 gives you more detailed information about the way that the study will be run.

Please take time to decide if you want to take part.

Part 1

1. Why are we doing this research?
This study is being done so that we can find out more about rare diseases in children and adults. These diseases include Wolfram syndrome, Alström syndrome, Bardet
Biedl syndrome and other rare diseases. This study aims to enroll children and adults with one of these rare diseases in an international register, to understand what causes these conditions. If we can find out more about these conditions, this may help us to develop specific treatments.

2. **Why have I been chosen?**
You have been approached because we think you have a rare disease. This study is planned to start in July 2011 and will include up to 600 children and adults with rare diseases.

3. **Do I have to take part?**
No. It is up to you to decide whether or not you take part. It is important to understand that taking part in this study is voluntary and you are free to stop taking part at any time. You don’t have to take part in this study to be looked after for your medical condition. Your doctor will explain the treatments that are available such as diet advice, medicines or insulin.

Before any tests are carried out, your doctor or nurse will explain the tests that will be done in this study and you will be asked to read this information sheet thoroughly. If you would like to take part and are happy with the explanations that your doctor or nurse has given, you will be asked to sign and date a form to confirm this. A copy of this information sheet and the signed and dated form will be given to you to keep.

4. **What will happen to me if I agree to take part?**
If you agree to take part, your doctor will tell the research team about your illness; any medicines you may be taking; and anything else from your medical notes that may be important to understanding your illness. This information will be entered into an anonymous, computer database: a ‘registry’. Anything that identifies you as an individual will not be on this database, and only the research team will have access to the database. You can have a copy of your medical information on the database if you like. The anonymised data will be stored in the registry for at least five years, and may be stored for longer than this.

You may also be asked to donate an extra blood sample. The amount of blood will be 10ml or 2 teaspoonfuls. You will not come to any harm as the body makes blood all the time. This sample will be sent to a laboratory to do or repeat the genetic test. Your blood sample contains DNA which is made up of genes. Looking at your DNA may help us to learn more about the genes that we think are linked to your condition.

We may also ask you to provide a small skin sample. The sample or ‘biopsy’ will be taken from your arm under local anaesthetic. The amount of skin that is taken is
about the same size as a match head. The skin grows back very fast.

The skin sample will contain cells. These cells can be used in the laboratory to grow more cells. The cells can be grown lots of times so can be used for lots of different research in the future without being used up.

It is important for you to know that you can take part in the registry (electronic database) part of the study without taking part in the skin biopsy or blood sample parts.

5. What are the side effects of the study and might I get any if I take part in the research?
Blood testing is part of normal assessment of your condition. The total amount of blood to be taken during the study is 10mls which is 2 teaspoonfuls. Every effort will be made to help you if you feel worried or anxious during a blood test. A special cream (a local anaesthetic) can be used to help numb your skin and you can have your parent or guardian with you during the test. If at any time you feel you cannot do the test as you are too worried, please just tell your study doctor/nurse.

6. What are the other possible disadvantages and risks of taking part?
Blood testing is a normal part of checking your illness. The amount of blood we will collect is about 2 teaspoonfuls. If you feel worried during the blood test, we will try to help you. The special numbing cream means the blood test will not hurt.

If you agree to give a skin biopsy we will use a local anaesthetic injection so that it doesn’t hurt. Sometimes (not very often) this wound may become red and sore. If this happens we will ask your parents to take you to see your doctor who will treat the problem.

7. What are the potential benefits of taking part?
There are no direct benefits in taking part. However, if we can put together the health information from a large number of patients, we may be able to identify times when we can make changes to the way we look after you. This may help you. We will also keep you regularly informed of new discoveries in the causes and treatment of your illness.

8. What happens when the research stops?
When the doctor has the genetic results from your blood test, he or she will tell them to you at your next outpatient visit; and discuss with you whether your treatment is affected by the result. When the research stops, your medical
care will continue just as before; but hopefully with new information to help your doctor look after you better.

9. **What if there is a problem?**
If there is anything you are worried about, talk to your doctor or nurse. There is more information about what to do if there is a problem in part 2.

10. **Will my personal data collected during the study be kept confidential?**
Yes. All information about taking part in this study will be kept confidential (private). More information regarding confidentiality may be found in part 2.

11. **Contact for further information**
If you require further information, please feel free to ask any questions you wish.

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Thank you for reading so far. If you are interested to find out more, please read Part 2 before making any decision.

12. **What happens if new information about my condition comes along?**
Sometimes new information comes along about the illness that is being studied whilst a study is still happening. You may feel differently about this study, and your doctor will discuss whether or not you still want to take part. If you decide to withdraw from the study, that is no problem, and your doctor will continue to provide care. If you continue, you may be asked to sign an updated consent form containing any new information.

Also, on receiving new information the study doctor might consider it to be in your best interests to take you out of the study. He or she will explain the reasons but continue looking after you. If the study is stopped for any other reason, we will tell you why, but your care will continue.
13. **What will happen if I don’t want to carry on with the study?**

It is important for you to understand that taking part in this study is **voluntary** and you are **free to withdraw at any time** without a reason. You do not have to take part in this study to be treated for your condition.

If you decide to stop taking part in the study, you will only need to inform your doctor and he or she will withdraw you from the study. This will not affect the care you receive. If you want to stop after you have given a blood or skin sample, you can choose either to have all your samples and data permanently destroyed; or anonymised so that no-one will ever be able to link your name to the tissue and/or data. Any research results that have already been done can’t be destroyed.

**What if there is a problem?**

If you have a concern about any aspect of this study, please speak to your doctor or one of the study doctors. We will do our best to answer these questions (see contact details in Part 1)

If you are still unhappy and wish to complain, your parents can find out how to do this at your hospital.

14. **Will anyone else know I am taking part in this study?**

Yes. If your parent or guardian has given consent for you to take part in this study and if you have signed the consent form, any of your medical records may be looked at by your doctor to collect health information for the study.

None of your identifiable data such as your name or address, will be put into the registry. Only your local doctor will have access to these details. Your data will be given a code number. Your doctor will keep a list of the code numbers and names and won’t share this list with the researchers. Any laboratory samples (e.g. blood samples) will have the code number on too.

In this way, the researchers will not know who the data and samples are from; but if they find something that may affect the way we need to treat you, they can pass the information to your doctor with your study code number. The doctor can then identify you and make sure you are offered the best available treatment.

15. **Involvement of your General Practitioner/Family Doctor (GP)**

We will tell your GP (and other doctors who are involved in your care) about you taking part in the study. We might also need to ask for more information from the GP about your medical history or medications you have taken. The GP may be told about your progress during the study.
16. **What will happen to any samples I give?**

Your samples will have all of your personal details removed from them (such as your name) so that they are only known by your registry unique identifier. Your samples will be stored and will be used in this project or for other research for your condition.

17. **Will any genetic tests be done?**

Yes. Your DNA will only be used to look at mistakes (mutations) in the genes for your condition. It will only be made available to scientists studying these conditions. In a few cases, the tests may find a result that will affect the way you need to be treated. If this happens, the results will be given to your doctor so that he or she can put you on the best possible treatment.

18. **What will happen to the results of the research study?**

Once the study is complete, the results will be published in a final report written. You can contact the study doctor if you would like to see the information that is published. Your name or personal details won't be written in any of the reports.

19. **Who is organizing and funding the research?**

The study is organized by Birmingham University and funded by the European Union.

20. **Who has reviewed this study?**

This study has been looked at by experts from the European Union and has also been reviewed by an Ethics Committee. It is the Ethics Committee's job to make sure that the study is safe and well designed.

The study will follow the UK guidelines for research to make sure that people taking part are kept safe.

If you wish to take part in the study you will be asked to sign the consent (agreement) form. However, you can only take part if your parent or guardian also consents to take part in the study. A copy of your signed consent form and this information sheet will be given to you to keep.

Thank you for taking the time to read this information sheet.