EURO-WABB Registry: An EU Rare Diabetes Registry
Parent or Guardian Information Sheet

My name is Dr Timothy Barrett, and I am working with your doctor. I am based at Birmingham Children’s Hospital, UK, and I look after people with Wolfram, Alström, Bardet Biedl and other rare diseases. I am the chief investigator for EURO-WABB (www.euro-wabb.org). EURO-WABB is a research study. The study involves the development of a European registry of children and adults who have been diagnosed with rare diseases such as 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 agree to your child taking part in the EURO-WABB research study.

Your child’s participation in this study is entirely voluntary. Before you decide if you want your child to take part, it is important that you understand why the research is being done and what it will involve. Please read the following information carefully and discuss it with your child, 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 your child 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?

Wolfram syndrome, Alström syndrome, Bardet Biedl syndrome and other syndromes are rare diseases with no specific treatment. The purpose of this research is to understand why people with these conditions become ill. This study aims to enroll children and adults with these rare diseases in an international registry, to obtain blood and tissue for genetic diagnosis and to understand the disease mechanisms. Given their rarity, the natural history of these diseases is still not well understood. Collecting patient data in this registry and learning about the progression of disease may speed up the development of clinical trials to test new treatments when they become available. Knowing more about these syndromes at the molecular level may also help to develop specific treatments for this disease.
2. Why has my child been chosen?
You have been approached because your doctor thinks your child may 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. You are being asked to take part in this study to understand your child’s condition better. We will do this by studying the genetic cause in a blood sample from your child, and by following your child’s progress over time. This will help us to understand what happens to people with rare diseases, and to offer future studies for new treatments to either halt or delay its progression.

3. Do I have to take part?
No. It is up to you to decide whether or not your child takes part. It is important to understand that taking part in this study is voluntary and you are free to withdraw your child at any time without a reason. This will not affect your child’s care in any way.

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

4. What will happen to my child if I agree to take part?
If you agree to take part, your doctor will ask you questions about your child’s illness. Your doctor may also add anything from your NHS medical notes that he or she thinks may help understand your child’s illness. Your doctor will then put this information on an anonymous database with the information from many other people with the same condition. You can have a copy of the information held about your child on the database if you like. You can also view your child’s data and add to the data if you wish. The anonymised data will be held on the registry for a minimum of five years, and may be stored for longer.

Your child may be asked to donate an extra blood sample. The amount of blood will be 10ml or 2 teaspoons. Your child 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. Any sample left over will be stored.

We may also ask your child to provide a small skin biopsy from his/her outer arm under local anaesthetic. If your child is under 2 years of age, he/she will not be approached to provide a tissue biopsy. The amount of skin taken is about the same size as a match head. The skin grows back again very fast. The skin biopsy is to make permanent cell lines from the skin cells. Cell lines are cells grown in a laboratory and have the same characteristics as the original sample cell. The cells are able to multiply, which means that they can be used for different research without being used up over time. Stored blood samples and cell lines will be used to study your disease and its complications over many years.

Both procedures will take place at the hospital. If the hospital already has these samples stored, we may ask your permission to use these samples for research.

It is important to understand that your child can take part in the registry without your child taking
part in the skin biopsy or blood sample parts of the study.

5. **What are the side effects of the study and will my child be at risk?**
There are no bad side effects to this study; the good effects are that this study will give us more information about your child’s illness and help to offer the most effective treatment.

6. **What are the other possible disadvantages and risks of taking part?**
Blood testing is part of normal assessment of your child’s illness. 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 the blood test. A special cream (a local anaesthetic) can be used to help numb the skin.

Skin biopsies may be taken from your child’s outer arm. A local anaesthetic (injection) will be used so that the procedure shouldn’t cause discomfort. After the procedure, you will be given advice about how to look after the skin break. Very rarely this skin break may become infected. If this happens, you should see your GP who will decide if any treatment is required.

7. **What are the potential benefits of taking part?**
If you have not yet had a genetic test for your condition, then this will be done as part of the study and you will receive the results. There are no direct benefits to you in taking part. However, if we can put together the clinical features from a large number of patients, we may be able to identify times when interventions may help your child. We will also keep you regularly informed of new discoveries in the causes and treatment of the syndrome.

8. **What happens when the research stops?**
When the doctor has the genetic results from your child’s blood test, he or she will tell them to you at your next outpatient visit; and discuss with you whether your child’s treatment is affected by the result. When the research stops, your child’s medical care will continue just as before; but hopefully with new information to help your doctor look after your child 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 child’s personal data collected during the study be kept confidential?**
Yes. All information about taking part in this study will be kept confidential. 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.

*Researcher’s name:* Prof Timothy Barrett  
*Address:* Diabetes Unit  
Birmingham Children’s Hospital  
Steelhouse Lane
Part 2

12. What happens if new information about my child’s 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 your child to take part. If you decide to withdraw your child 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 child’s best interests to take your child out of the study. He or she will explain the reasons but continue looking after your child. If the study is stopped for any other reason, we will tell you why, but your medical care will continue.

13. What will happen if I don’t want my child 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 your child at any time without a reason. You do not have to take part in this study for your child to be treated for his/her 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 your child from the study. This will not affect your child’s standard of care or the benefits to which your child is entitled. Should you wish to stop after your child has given a blood or skin sample, you can choose either to have all your tissue samples and data permanently destroyed; or anonymised so that no-one will ever be able to link your child’s name to the tissue and/or data. Any research results already generated from your child’s blood sample and or clinical data cannot be destroyed or recalled.

What if there is a problem?
Complaints:
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 formally, you can do this using the NHS complaints procedure. You can ask how to do this at your hospital.

Harm:
If you have an accident while having the blood sample taken on NHS premises, the NHS has the indemnity for this study. We will make sure you are closely supervised while having the blood test in the hospital.
14. Will anyone else know my child is taking part in this study?

Yes. If you have given your consent for your child to take part in this study, any of your child’s medical records may be looked at by your doctor to collect health information for the study. Also, people from the regulatory authorities (such as the Department of Health) may look at your medical notes to check that the study is being carried out properly.

None of your child’s personal data, such as name and contact details, will be uploaded onto the registry. Only non-personal data will be put onto the registry. Your child’s blood data will be assigned a code number. A master list linking the code number and your child’s identity will be kept separate from the research data. The master list will be kept in a locked file and your child’s data will only then be identified by a subject number (this is a number you get you join the study). Any laboratory samples (eg blood samples) will be identified in the same way. None of your child’s personal details will be used in any presentations and publications as required by local/regional/national regulations. The information about your child and his/her blood and tissue samples are confidential and will not have your child’s name and address on them. The information is also anonymous but has a unique identifier (the subject number referred to above). In this way the researchers analysing your child’s data and blood or tissue sample will not know who they are from; but if they find something that may affect the way we need to treat your child, they can pass the information to me with your unique identifier. I can then identify you to pass the information back to your local doctor and make sure your child is offered the best available treatment.

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

We will tell your GP (and other specialist doctors who are involved in your healthcare) about your child taking part in the study. We might also need to ask for more information from your GP about your child’s medical history/medications he/she may have taken and the GP may be told about your child’s progress during the study.

16. What will happen to any samples my child gives?

With your consent, any samples will be anonymised so that they are only known by your registry unique identifier. All UK samples will be stored in Birmingham in a licensed bio-repository. Samples will be stored for as long as they have research value and may be shared with other approved research workers in the UK or abroad. This means that, with your agreement, the samples will be stored for a minimum of three years, and may be stored for longer.

Blood samples will be used for genetic analysis of genes associated with your child’s disease.

It is possible that the samples you donate may be used in future, as yet unidentified studies that relate to your child’s disease or its complications.

Tissue samples, e.g. from a skin biopsy, can be used to generate stem cells which carry one of the genes associated with your child’s disease. These stem cells may then be used to produce cells representative of specific organs (such as heart kidney etc).

17. Will any genetic tests be done?
Yes. Your child’s DNA will only be used to look at mistakes (mutations) in the genes related to his or her condition. It will only be made available to scientists studying these conditions. In a few cases, the genetic tests may find a result that will affect the way your child is treated. If this happens, the results will be passed back to your child’s Consultant to put him/her 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 and a final report written. You should contact the study doctor if you would like to get copies of any information that is published. You will not be identified in any reports or publications.

19. Who is organizing and funding the research?
The study is organized by The University of Birmingham, UK, and funded by the European Union Directorate General for Health and Consumers.

20. Who has reviewed this study?
This study has been reviewed by independent expert peer reviewers for The European Union Directorate General for Health and Consumers.

The study will be run in accordance with all suitable guidelines aimed at ensuring proper conduct and safety of anyone taking part, including the Guidelines on Good Clinical Practice and the Declaration of Helsinki.

If you wish to take part in the study you will be asked to sign the consent (agreement) form overleaf. 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.