



Study

<http://goldstudy.cimr.cam.ac.uk>

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Genetics of Learning Disability (GOLD) Study INFORMATION SHEET FOR FAMILIES PARTICIPATING IN THE GOLD STUDY

Introduction

You are being invited to take part in a research project to study how often genes cause learning disability. Before you decide you may wish to know why the research is being done and what it will involve. Please take the time to read the following information carefully and discuss it with others if you wish. Ask us if there is anything that is not clear or if you would like more information. Take time to decide whether or not you wish to take part. Thank you for reading this.

We would like to invite you and your family to participate in a research project to study how often genes cause learning disability.

1 in 50 children in the population has significant learning disability and 1 in 200 has severe problems. Parents who have a child with learning disability usually want the answer to two questions:

1. Why does my child have learning disability?
2. Will my next child have learning disability?

For the majority of such children no one else in the family has learning disability and the routine tests rarely reveal an obvious cause.

Research

Scientific research into the genetic causes of learning disability in the last 10 years has found at least 65 possible genetic locations for learning disability and, in the last 2 years, isolated the first 10 of these genes. Identification of mistakes in one of these genes in a child with learning disability proves that the cause is genetic and, as a consequence, that there would be a precise 1 in 4 chance for any further children in the family to have similar learning disability.

We are collecting blood and/or saliva samples from families who have one or more children with learning disability to find out whether they carry a mistake in a gene that is responsible for the learning disability. This project is funded by the medical research charity, Action Medical Research.

We are collaborating with all the clinical genetics departments in the UK and also with colleagues in Australia, USA and Europe. Families that have previously been seen by their local genetics services are being invited to take part if they fit the

appropriate inclusion criteria (for more information visit our website at <http://goldstudy.cimr.cam.ac.uk>). We aim to recruit up to 2000 families from the UK.

Your local genetics department has identified that your family may be eligible to take part in the study although the research team do not know who you are as yet. Information about you will not be released to us until after you have given your consent for this information to be released and that you wish take part in the study.

You and your family are under no obligation to participate in the study and if you would prefer not to then we will accept your decision without question and will continue to treat you as before. If you do decide to take part you will be given this information sheet to keep and you will be asked to sign a consent form. If you decide to take part you are still free to withdraw at any time during the study without needing to give a reason and without it affecting the medical care of you or your child.

All the information which is collected about you and your family during the course of the research will be kept strictly confidential. If you withdraw from the study at any time all the information we have stored will be kept to the end of the study but your samples and data will be withdrawn from the study and any further analysis.

If you decide to take part in the research project the next step is to

- tell your local geneticist that you wish to take part
- keep a copy of this information sheet
- sign 3 copies of the consent form and give 2 copies to the geneticist, who will forward one copy to me and one will be placed in your local hospital notes. The 3rd copy is for you to keep as a record.

If there is sufficient DNA already stored on all your family members no further contact will be needed at present. If your family is eligible but more DNA is needed for the study the following will happen:

- someone from either your local genetics team or the research team in Cambridge will contact you to arrange for the collection of a blood/saliva sample.
- A saliva collection kit will be sent to you in the post with full instructions on its use and how to return it to us free of charge.

Blood Samples

Blood samples will be taken or sent to the laboratory in Cambridge where a cell line will be made on some of them so as to avoid taking further blood samples if we need more DNA. The blood sample that is required is 10mls or 2 teaspoonfuls of blood. This will be taken from the arm using the same technique that is used when you go to the doctors for a routine blood test. It is painful as the needle enters the skin but short lived and can be associated with slight bruising at the needle entry point.

Making a cell line involves using a portion of the blood sample taken and making the blood cells grow continuously by adding a virus and growing the cells in special culture medium. This means that should we require a further blood sample from someone the cell line can be used instead and thus avoids additional blood samples being taken in the future. The rest of the sample will be used to extract DNA and will be stored in Cambridge in the research laboratory and analysed.

Saliva Samples

The saliva sample required is 1-2mls of saliva. A self collection kit will be provided together with full instructions on how to collect the sample and how to post it back to us. This is a non-invasive procedure with no risk or discomfort and will be a better experience than having a blood sample taken. In the event that the person providing the sample cannot spit the required 1-2mls, for example infants and young children, saliva sponges may be used to soak up saliva from the cheek pouches. Again this is non-invasive and has no risk or discomfort. If it is necessary to use saliva sponges to collect the sample then we will contact you directly to discuss the best way of doing this.

Samples we collect will not be used in any other research projects unless new consent is obtained from the participating families.

Complaints

If you are harmed by taking part in this research project, there are no special compensation arrangements. If you are harmed due to someone's negligence, then you may have grounds for a legal action but you may have to pay for it. Regardless of this if you wish to complain about the way you have been treated during the course of this study, the normal Health Service complaints mechanism may be available to you.

Results

The results of the study will help to develop a routine service to test children with learning disability and provide information to parents and other family members. As this is an ambitious project we do not expect to get results immediately and it may take up to 5 years to find a result.

Unfortunately we do not expect to get a result for every family taking part. However, we do undertake to feedback the general results of the study findings to all participating families via their local genetics department. Where we do get a result that is of direct benefit to a family we will inform their local clinician and ask them to communicate the findings directly to the family. All research findings will need to be verified in a NHS accredited molecular genetics laboratory before being used in routine clinical practice.

Contacts

Finally, I would like to thank you if you feel able to take part in this study. Thank you also for your time in reading about the study if you are not able to participate.

If you have any queries about this study or your participation in it please do not hesitate to contact:

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