



## **Title: Molecular Genetics of human birth defects**

### **INFORMATION FOR PARTICIPANTS 8-14 years.**

We are working all the time to see if we can try to help young people who have an inherited genetic disorder, which is a problem that is thought to be caused by faulty genes but which may also appear 'out of the blue'. We are working on a variety of conditions like this, that affect the formation of the brain, heart, limbs, kidneys, skin, bones, and immune system. Therefore, we ask your permission to take some blood when you are in hospital, and have a look at your genes (DNA). Take time to decide if you want to say YES or NO to this. Please read this information or have someone to read it for you. Don't worry if you don't understand it straight away. Your parents have also been told about this, and you can ask them to help you understand.

#### **1) Why are we doing this?**

We want to see if we can understand the reasons why some children are born with a problem that affects their normal development, while others are not. In the future, better understanding of this will allow us to decide which young people need treatment.

#### **2) What will be different for you?**

We will be asking your parents and you some questions when you come to hospital. We will also be asking you to have a blood test and will take a small blood sample. We can put some numbing cream on your arm so that you do not feel any pain while blood test is done. We may alternatively ask for a saliva sample. The test can be done at any time that is convenient to you and your family. We may ask for a urine sample. The doctor might also ask to take a sample from your nose using a small brush. This might feel uncomfortable but is not dangerous. We may also ask to take 4-5 hairs from your head, or for a small skin sample, which involves removal of a circular piece of skin (about the size of the blunt end of a pencil) after the skin has been numbed with a local anaesthetic to minimise discomfort. This might feel uncomfortable but is not dangerous. It may lead to a small scar, therefore is performed on less exposed skin areas.

#### **3) Why are we asking you?**

If you are a young person who might have an inherited disease, we are asking that you participate so we can understand the disease affecting you and others more clearly.

#### **4) Do I have to take part?**

No. It is up to you and your parents to decide. If you decide you don't want to, that's absolutely fine. The doctors and nurses will look after you as best as they can anyway.

#### **5) What about the results of the tests?**

The doctors taking care of you will discuss the results with you and your parents if you wish.

#### **6) Who will know about me and my results?**

The doctors organizing the tests will know your results and will share these with other scientists and doctors around the world, but they will not tell these other people your name. We will also send your samples to other doctors and scientists, but these people will not know that the samples came from you.

Your results might also be printed in a magazine called a scientific journal. Doctors and nurses read these to learn of new medical discoveries. Your name and details will always be taken off so that nobody knows whose results they are.

If you say YES, we will let your GP know that you are taking part in the study.

#### **7) Who can I speak to if I have any questions?**

You can speak to your parents who have also been given information about this project. You can also speak to the doctors or nurses on the ward.

You and your parents can always speak to the doctors involved in this work if you have any more questions. Your parents also have some further contact details of people to speak to if they have any complaints or worries.

Please do not hesitate to contact him/her if you have any questions or comments about the study. You can write/email to:

Prof. Philip Beales:	p.beales@ucl.ac.uk
Dr. Hannah Mitchison	h.mitchison@ucl.ac.uk
Prof. Peter Scambler	p.scambler@ucl.ac.uk
Dr. Miriam Schmidts	m.schmidts.10@ucl.ac.uk
Dr. Paul Brogan	p.brogan@ucl.ac.uk

**At the ICH, 30 Guilford Street, London WC1N 1EH.**

Or telephone 020 7242 9789 ICH switchboard.