

Version 5, 20<sup>th</sup> June 2017

## **Genetics of Growth Hormone Insensitivity (GHI) (Primary IGF-1 Deficiency, PIGFD)**

### **Information sheet for patients aged 12-16 yr**

This study aims to find out why you are small. Growth hormone insensitivity (GHI) or primary IGF-1 deficiency (PIGFD) is a condition that causes poor growth and leads to people being short. We are doing a study to look at genes that might cause problems with growth. We would like you to agree to allow us to take a blood sample from you.

#### **Why have I been invited?**

You have been invited because you may have a condition called growth hormone insensitivity (GHI), this means that you are not growing properly. Several genes have been found to cause the growth problem in patients with GHI but the cause is not known in many cases. Investigating your genes may help us to understand more about how GHI happens, and hopefully it will lead to new and better treatments to help other patients in the future. Your blood sample will help us to see if there is something in your genes (DNA) in your body that is causing the problem.

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

No-it is up to you to decide whether or not to take part. You will be asked on another form if you would like to be part of these studies. You don't have to do this and you can change your mind at any time. If you decide not to take part, this will not affect the care you receive.

#### **What will happen if I take part?**

We will need to take one small sample of blood (this means having a

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needle prick to get the blood), and we will carefully look at your genes. Very rarely, we will also ask for a small sample of your skin to get more information about your genes. To do this, your skin is cleaned and anaesthetic is put on your skin to make it numb. When the skin is numb, a small circle of skin (3-4mm, less than a quarter the size of your little fingernail) is taken using a tool a bit like a hole punch. You might see a little bit of bleeding but stitches are not usually needed. You need to keep the area dry for a day and the dressing can be taken off after about 2 days. It is up to you to decide whether or not to give us a skin sample and you should tell us if you want to or not on the form.

### **What are the possible benefits of taking part?**

We cannot promise the study will help you but the information we get might help treat young people with growth hormone insensitivity (GHI) with better medicines in the future.

### **Will my taking part in this study be kept confidential?**

Only your parents, the doctor and nurses looking after you and the people doing the research studies will know that you are taking part. We will also let your family doctor (GP) know.

### **What will be done with the information and blood used in the research?**

All information and samples that are collected during this research study will be used to learn more about growth problems. The samples will be kept for 20 years in a tissue bank for further use in future ethically approved research projects

### **Did anyone else check the study is alright to do?**

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A group of people called the Ethics Committee have checked that the study is OK to do. The Cambridge East Research Ethics Committee has approved the research study. We are also asking your parents permission.

**Who can I contact to find out more about this study?**

If you have any questions please contact the following:

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