Barts Health NHS Trust

Barts and The London School of Medicine and Dentistry

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Genetics Factors Affecting the Timing of Puberty

Information sheet for adult patients

What is the purpose of this study?
This study aims to identify genes that play a part in the timing of puberty. We are recruiting patients who are affected by a disorder of puberty, where the timing of puberty is significantly different to normal. These conditions are often passed down in families from parents to children and therefore it is very likely that there is a genetic cause for abnormalities of pubertal timing. We are using new genetic techniques to look for gene mutations (changes or problems with a gene) that cause abnormal pubertal timing. The current understanding of these conditions is limited and we are always looking to find better types of treatment. The best way to develop new treatments is to understand how the problem starts in the first place, and we are actively involved in researching these disorders.

Why have I been invited?
You have been invited because you have (had) a disorder of pubertal timing. Studying your genes may help us to identify possible genetic causes of disordered pubertal timing. Investigating your genes may help us to understand more about how these conditions develop and hopefully it will lead to new and better treatments in the future. We would therefore like to look in more detail at your DNA to search for genetic causes of abnormal pubertal timing.

What genetic studies are done?
We analyse the genetic information in your blood to look for changes (mutations) in known genes or new genes that may cause abnormal pubertal timing. We then compare these genetic changes to those in other individuals (both affected by disorders of pubertal timing and their unaffected family members) to try to identify which are the important genes causing abnormal pubertal timing. We then carry out further tests to try to understand the role these genes have in the body and how these gene mutations affect the timing of puberty. These genetic tests will not affect your medical treatment in any way.

Do I have to take part?
It is up to you to decide whether or not to take part because participation in the research study is completely voluntary. If you do agree to take part you will be given this information sheet to keep. You are under no obligation to participate in any part of this study, and you may withdraw at any time without it affecting your normal medical care in any way.

What will happen if I take part?
We would like to take a single sample of blood **(this means having a needle put into a vein in your arm in order get the blood)**, about a tablespoonful (15 ml), and from this we will remove the cells and isolate the chemicals carrying the genetic information (DNA and RNA). We will study these to see if there are any abnormalities in their sequences.

Will my taking part in this study be kept confidential?
Only the medical team taking care of you and the individuals doing the research (Dr. Sasha Howard and members of her research team from Queen Mary University of London) will have access to your personal information and the data collected during the study. Once you agree to participate in the study and sign the consent form, one of the members of the medical team will take a blood or DNA sample and some information. This information includes your birth weight, weight, height, family

history, timing of puberty and results of any relevant tests that you have had done. All the information that is collected about you during the course of the study will be kept strictly confidential. We will not share your clinical data or genetic results with anyone other than your consultant. Your clinical details and results will be kept on a computer database. This information will only be used for for research purposes, kept secure and is accessible only via strict password protection. We will notify your GP that you are taking part in the study unless you tell us not to. If you consent to take part in the research the people conducting the study will abide by the Data Protection Act 1998, and the rights you have under this Act. For the purposes of monitoring of the quality of the research, authorised persons such as regulatory authorities and persons from the NHS Trust may also have access to relevant sections of your medical notes and data collected during the study. You will be asked on the consent form whether you agree to your clinical information being shared with the research team and other authorised persons.

Will I be told the results?
The results of the genetic studies will not be fed back to patients or their families. These results will be limited to problems with the timing of puberty. Although the study is likely to have overall benefit for patients with problems with pubertal timing these benefits are likely to take up to 5 years to have effect. The study will contribute to the understanding of the genetics and biology underlying abnormal timing of puberty but is not designed to have individual patient benefit at this stage.

What will be done with the information and blood used in the research?
All clinical information and samples that are collected during the course of this research will be used to gain further information about disorders of puberty. The samples and the data will be stored securely. Only the study doctors will have access to your identity, the study data and study samples. The samples will be kept for future ethically approved research projects. Your clinical data will be securely destroyed at the end of the research studies.

What happens if there is a problem?
Queen Mary University of London has agreed that if you are harmed as a result of your participation in the study, you will be compensated, provided that, on the balance of probabilities, an injury was caused as a direct result of the intervention or procedures you received during the course of the study. These special compensation arrangements apply where an injury is caused to you that would not have occurred if you were not in the study. These arrangements do not affect your right to pursue a claim through legal action. Please contact Patient Advice and Liaison Service (PALS) if you have any concerns regarding the care you have received, or as an initial point of contact if you have a complaint (telephone 020 359 42040 / 42050, or email PALS@bartsandthelondon.nhs.uk). You can also visit PALS at the Royal London Hospital.

What will happen to the results of this study?
The results of this research will be disseminated by presentations at scientific meetings and published in scientific journals. No identifiable information will be included and all data will be anonymised before presentation or publication.

Who is funding the research?
Barts and the London Charity and the Academy of Finland are currently meeting the cost of the studies. No payment is being given to the doctors or researchers for including you in this study. The study is being conducted as part of an educational project (PhD).

Who has reviewed the research?

The London-Chelsea Research Ethics Committee has approved the research proposal.

Who can I contact to find out more about this study?
If you have any questions please contact the following:

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Thank you for considering taking part in this important study. We would very much appreciate it if you were able to help us.