Screening for Stickler Syndrome in children diagnosed with Perthes Disease

Information for Participants and Parents

What is the purpose of this study?
The aim of this study is to investigate the occurrence of Stickler syndrome, a rare genetic condition, amongst paediatric patients being diagnosed and treated for Perthes disease. Identification of these patients would allow the offer of earlier treatment, preventing retinal detachment and potential blindness. Whilst most patients with Perthes disease will not have Stickler syndrome, this study aims to identify any patients with this condition using a screening tool and a genetic test.

Why has my child been chosen?
Your child has been referred to the paediatric orthopaedic team in Alder Hey Children's Hospital for Perthes disease. All patients in this clinic at this time are being offered this screening assessment for Stickler syndrome.

Who is organising the study?
The study is being organised by Dr Robert Smyth, paediatric trainee and Addenbrookes Charitable Trust Clinical Research Fellow. This is under the supervision of Mr Martin Snead, Consultant Eye Surgeon at Addenbrooke's NHS Trust and Vitreoretinal Research Group lead at the Van Geest Brain Repair Centre, University of Cambridge. It is also supervised by Dr Peter Bale, Paediatric Rheumatology Consultant and Paediatric Musculoskeletal lead at the NHS England Stickler Syndrome Service at Addenbrooke's NHS Trust.

What will happen to my child if I take part and what do I have to do?
We will ask you/your child some questions from our screening tool about conditions associated with Stickler syndrome. This should take 5-10 minutes, and we may also ask to briefly look at your child's mouth and joints. We will also ask to collect a blood sample from your child for genetic testing for Stickler syndrome. Any genetic tests performed would be specifically for Stickler syndrome and not done on any other part of your child's DNA.

What are the possible risks/side effects of taking part?
As we would require a single sample of blood from your child for a genetic test, there will be the normal risks of bruising and distress associated with any other blood test.

What are the possible benefits of taking part?
Stickler syndrome is a rare but important condition that runs in families and can result in retinal detachment and sight loss. If your child is identified as having Stickler syndrome, then we will write to your GP to arrange referral for urgent assessment at the Highly Specialised Service for Stickler Syndrome in Cambridge. This is a national service, free at

the point of use, and offers high quality care from several specialist teams for patients with Stickler syndrome. If referred, your child will have their genetic test repeated to confirm the diagnosis, and the rest of the immediate family will also be assessed, as this is a genetic condition. This study will also benefit future families by helping us to identify patients with Stickler syndrome earlier and give potential sight-saving prophylactic treatment.

What if new information becomes available?
Any new information will be communicated to participants and their GP.

Confidentiality – who will have access to the data?
Participant confidentiality is paramount. Access to clinical information will be restricted to the principal investigator in charge of the study (Dr Robert Smyth) and his named research associates who are also medically qualified.

Will my GP be informed?
Yes. Your GP will be informed by letter with details of the study, and if any genetic testing is positive. The paediatric orthopaedic team currently managing your Perthes disease will also be informed so that they can ensure your ongoing treatment is suitable.

What will happen to the study results?
The results of the research will be submitted for publication in scientific journals. All participant information will be anonymised. Results of genetic analysis will be maintained to allow future anonymous analysis in related projects. Your child's blood sample will also be stored in order to allow future tests to be done if new information becomes available. This will only be performed as part of ethically approved research and will not be used for any commercial purposes. Any clinically relevant results from future research would be communicated to you/your child as above.

What happens if I do not wish to take part, or wish to withdraw during the study?
If you do not wish to take part, you do not have to give any reasons for your decision, and this will not affect any treatment you may require as part of your personal care. Similarly, if you decide to take part and then wish to withdraw from the study you are free to do so at any time and you do not need to give a reason.

Local contact for information
Thank you for reading this and considering our study. If you have any further questions at any time I can be contacted via the vitreoretinal centre in Cambridge on 01223 348842, or directly by e-mail at rs815@cam.ac.uk

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