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. This part of our research is designed to test our screening tool's ability to differentiate children with or without Stickler syndrome.

Why has my child been chosen?
Your child has been chosen as they are aged between 4-10 years with or without Stickler syndrome and have not previously been diagnosed with Perthes disease or another connective tissue disorder.

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 they 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. You will not be required to provide any further information. We may contact you again in the future with other questions if necessary.

What are the possible risks/side effects of taking part?
Since this research is predominantly survey based, we do not anticipate any risks or side effects associated with your child's participation.

What are the possible benefits of taking part?
The study is designed to benefit future patients and families by helping us to identify patients with Stickler syndrome earlier and thus provide potentially sight-saving prophylactic treatment.

What if new information becomes available?

Any new information that becomes available 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?

No. Your GP will only be informed by letter with details of the study if there is new data that becomes available that is clinically relevant.

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.

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

Dr Robert Smyth MB BChir MA with Hons, ACT Clinical Research Fellow and Paediatric Trainee, Addenbrooke's Hospital, Cambridge, CB2 0QQ