



umcg

Information about research into sloping shoulders in CHARGE syndrome

With this flyer we would like to provide more information on the research about sloping shoulders that we will be conducting at the 12th International CHARGE Syndrome Conference.

Why do we want to investigate sloping shoulders in CHARGE syndrome?

We have noticed in our CHARGE clinic that many children with CHARGE syndrome have sloping shoulders. For many this does not pose a problem. Some, however, develop problems with lifting their arms above their head. In true CHARGE form, they usually find a way around it! However, we have found that a very small group of CHARGE children go on to develop more general muscle issues, such as muscle weakness in their arms or legs. That is why we want to investigate whether having CHARGE syndrome may have an effect on the muscles itself. To do this, we will perform muscle ultrasounds in CHARGE patients with and without sloping shoulders.

Who can participate?

- Children with definite CHARGE syndrome:
 - Children with a proven CHD7 mutation
 - Children with a clinically 'definite' CHARGE syndrome diagnosis, who have not had CHD7 diagnostics done. These children can be included if two experts independently agree with the clinical diagnosis
- Children **with or without** sloping shoulders
- Able to sit reasonably still during the ultrasound.

What do we ask of you?

How will the research be conducted?

We will perform dynamometry and ultrasound examinations. *Dynamometry* is the measurement of muscle strength. To test this, the child is asked to exert force on a hand-held device (a dynamometer). It is a simple and non-painful test. Most children enjoy showing off their muscles! *Ultrasound* uses sound waves to visualize tissues, such as muscles. It is a simple and safe examination that is not painful. The ultrasound gel can feel a little cold. We will examine the muscles in your child's arm and leg (on one side). The only requirement is that your child needs to be able to sit reasonably still for about 15 to 20 minutes. We will also take *photographs* of your child, to document the shape of his or her shoulders

How long will it take? Estimated time: 30-45 minutes in total, including time for introductions and questions.

Informed consent

We will ask you to sign an Informed Consent form. By signing this form you show that you understand what the study is for and agree to take part.

What about risks, privacy and confidentiality?

There are no risks in taking part in this study. We will keep your personal information private and confidential. Our database is stored on a secured hospital server with restricted access. No information will be given to others without your permission. The overall results of this study may be shown at professional or patient group meetings or published in journals to inform other doctors and health care professionals. We will endeavor to keep your identity private in any publication or presentation. If we are interested in publishing photographs of your child, we will always ask you for your permission separately.

How can you reach us?

If you are interested in participating in our study, please contact us at the email address below. If possible, we would like to schedule appointments beforehand – **send us an email**. You are also welcome to meet us at the conference to plan an appointment at that time. We will be wearing buttons with 'Ask me about sloping shoulders'!

Drs. Christa de Geus
Clinical geneticist in training & PhD student
University Medical Center Groningen
The Netherlands
CHARGE@umcg.nl

This research is supervised by:
Professor C.M.A. van Ravenswaaij-Arts Clinical geneticist & CHARGE researcher

Ask me about



Sloping shoulders