



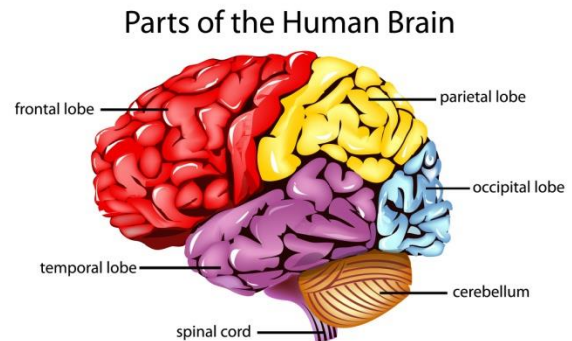
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## Information about “the role of the brain in balance in CHARGE syndrome”

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

### Why do we want to study the role of the brain in balance?

Children with CHARGE syndrome often have poor balance. We know that abnormalities of the semicircular canals, which are located in the inner ear, are one reason for this. However, maintaining balance is a complex process in which the brain plays an important role, too. The part of the brain that is most important for balance is the cerebellum. The cerebellum (“small brain”) is located at the back and the base of the brain (see figure). When a person has cerebellar problems, they will



often have difficulties with balance and coordination. A person may walk with a staggering gait, slur their speech or be imprecise in their movements.

This combination of problems is called ‘ataxia’. We recently found that approximately 50% of CHARGE patients have cerebellar anomalies on their MRIs. Because of the role of the cerebellum in regulating balance, we wondered whether cerebellum anomalies could be an extra cause of balance problems in CHARGE syndrome and whether CHARGE patients may have more ‘ataxia symptoms’. To determine this, we are conducting a study in which we are evaluating the presence of balance and ataxia in CHARGE patients.

### Who can participate?

- Children with 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
- Age: between 4 and 18 years old, with developmental age: over 4 years old
- Able to perform SARA and Berg Balance scales. These scales involve a series of tasks (for instance: standing still, walking, walking in tandem). See below for further information.

We are especially interested in children who have had brain MRIs which can be requested for review (preferred but not required).

### What do we ask of you?

How will the research be conducted?

We will perform two tests that measure balance and coordination: the 'Berg Balance scale' and the 'SARA score for the assessment and rating of ataxia'. In these test children are asked to show how well they can perform a number of tasks. There are easier tasks, such as sitting up, and more difficult ones, such as walking in tandem or standing on one leg. The examination will be videotaped.

To participate, children should be able to understand simple instructions. Don't worry if your child cannot yet do some of the more difficult tasks: the difficulty is purposefully graded to allow evaluation of children of different ages and abilities. If your child communicates with sign language, we would appreciate your help with interpretation.

How long will it take?

Estimated time: the SARA and Berg scores take about 15 minutes to complete. To allow time for introductions and questions we estimate a total duration of 30 minutes.

MRI studies. If available, we will ask your permission to request copies of MRI studies that were done in the past. The images will be reviewed by a specialized pediatric neuroradiologist. You will be sent a copy of the review.

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 keep your identity private in any publication or presentation.

### **How can you reach us?**

If you are interested in participating in our study, you can 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. We will be wearing buttons with 'Ask me about balance'!

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