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Clinical, MRI and molecular investigations in people with a variation on chromosome 16p11.2
Information sheet for participants in the 16p11.2 study.

Dear Madam, Sir,

We would like you to participate in the present study because a genetic variation on chromosome 16p11.2 has been identified in yourself or your child. This variation may be responsible of delay in speech or development and learning difficulties, as well as a tendency to put on weight or lose some abnormally. This study will help us understand how this variation is associated with these different manifestations. There are also people carrying this variation who never present clinical manifestations and we want to understand why.

Course of the study:

To participate, you need to be older than 3 and carry this variation on chromosome 16p11.2, or be related to a person carrying it. All participants under 18 must be accompanied by a parent or legal guardian.

Family history

We will retrace your family history as some of your relatives might also carry this variant.

Neuropsychological evaluation

We will assess language skills, memory and attention span. The duration of this evaluation is between 60 minutes and 2 and half hours, depending on the age of the participant (shorter evaluation for children).

Behavioural evaluation

This evaluation will require 2 sessions (approximately 1 hour each) during which questionnaires as well as a structured interview will be performed. The evaluation in children will be adapted according to their age.

Blood sample

One blood draw in several tubes will be performed for the following analyses :

Genetic analysis for participating carriers (1 tube). This will help to determine precisely the size of the genetic variation on chromosome 16p11.2 which has been previously identified in your child or yourself.
Cell line for all participants (2 tubes) : The “cell lines” are established from white blood cells. These cells will multiply in the lab and will constitute a reusable sample. With this procedure, we avoid taking repeated blood tests. These cell lines will be stored under an anonymous code, for as long as 25 years in a freezer (-140 degrees C) with a controlled access. They will be destroyed after 25 years. These cells will only be used for this study.
Analysis of gene expression levels for all participants performed in cell lines. We will study the effect of this genetic variation on the expression of the genes in this chromosomal region.

Brain MRI

For participants older than 6 years of age, a magnetic resonance image scan will help us understand how the variation on the chromosome 16p11.2 may affect brain function. The MRI scanner works with a large magnet so you will be asked to remove all metal objects, such as rings, earrings, bracelets, belts etc. This procedure is perfectly safe but it is not possible for people with metallic prosthetics or electrical devices such as a pacemaker. We will take time to go through these exclusion criteria with you beforehand.
During the scan session, which lasts about 1 hour, you will hear different noises. First, you will lay motionless. If you wish, you can watch short movies without the sound. Then we will ask you to watch pictures and press some buttons according to different tasks that we will have discussed with you previously, outside of the scanner. The young participants may have a shorter session (30 min) where they have nothing to do. This examination can be stopped at any time you want.

Stool analyses

Intestinal bacteria will be studied on stool samples and will allow to better understand links with weight loss and gain.

Risks / benefits

Studying the effects of this variation on chromosome 16p11.2 may help prevent significant weight gain by early dietary support. Diagnosing specific language impairments can also help initiate early interventions such as speech therapy. During the course of this study, if evaluations show that you or your child would benefit from additional interventions, we will discuss it with you and inform your general physician.

Confidentiality

Your medical and genetic data will be securely archived. All data can eventually be sent to you or your GP. Your DNA extracted from the blood sample will be kept anonymously in the medical genetics department. The cells lines can be kept anonymously for a maximum of 25 years in liquid nitrogen or freezers (-140 degrees C) with strictly controlled access by Dr. Reymond from University of Lausanne. You can access or correct your medical data by contacting the

investigator of the study. You can as well, at any time, get your file back or ask for its destruction. The results of this study can be published in scientific journals but all data published remain anonymous.

Remuneration

The medical examinations in this study are completely free of charge. We will pay for your transportation, accommodation and food.

Voluntary participation

This protocol was reviewed and approved by the institutional review board (ethics committee) in April 2011. Your participation in this study is on a voluntary basis. You can, at any time, withdraw from the study without any justification. In case of withdrawal your patient and research participant rights are unchanged and we will remain available for any questions. At any time you can request the destruction of your DNA sample or cell lines by contacting the principal investigator.
In addition, it is important to keep in mind that it is not necessary to take part in all part of the study (e.g. you can only participate in the MRI part or only in the neuropsychological assessment part).

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