# STUDY OF THE GENETICS OF CHILDHOOD SPEECH/LANGUAGE DISORDERS

# FAMILY INFORMATION SHEET

## What is the goal of this study?

Recently, we found that having a duplication (or extra copy) of a small portion of chromosome 7 is associated with severe speech impairment and expressive language delay. This duplication has only been reported in a very few children, and so we are initiating a study to find out whether more children with speech/language impairment carry this duplication on chromosome 7. This study aims to test around 500 children with speech/language delay for the duplication.

## What specific set of characteristics is this study looking for in a child?

1. Speech/articulation delay and/or apraxia
   - Unclear speech that is difficult for others to understand (especially strangers)
2. Expressive language delay
   - May only be able to speak a small number of words
   - Uses gestures, signs, pantomime and/or drawing to communicate
   - Scores on expressive language tests below those on receptive language (comprehension) tests
3. May have mild developmental/intellectual delay (not always present)

## What does this study involve?

Participation in the study will require two or three things:

***1. A saliva sample from the child***

The saliva sample is collected by parents in the home by having the child spit in a tube. If the child is unable to spit, a cheek swab can be taken. The saliva sample is sent to Toronto where we will extract DNA for analysis of chromosome 7. If you decide to enroll your child in this study, we will send you a kit with detailed instructions on saliva collection and postage-paid shipment to the laboratory.

***2. A consent form signed by the child's parent or guardian***

This form is available from the contacts listed at the end of this pamphlet. The consent form outlines the study, what will happen to your child's saliva sample, and any potential risks or benefits associated with participation. This study has been reviewed by the University of Toronto Research Ethics Board and the Hospital for Sick Children Research Ethics Board, and the consent form has been approved by both of them.

***3. A summary of the child's performance on standardized speech and language assessments***

If your child is found to have a chromosome 7 duplication, we will ask you to share information (like reports or test results) about your child's speech and language skills. We will ask for your permission to speak with your child's Speech-Language Pathologist to

gather more detailed information. This will help us to see whether there are any specific characteristics shared by children who have a chromosome 7 duplication.

## Why is this study important?

Although scientists have shown that speech/language impairment has a strong genetic basis, only one gene has been linked to a speech/language disorder, and this only in a very few families. The study described here aims to determine whether duplication of a specific region of chromosome 7 is a *common* cause of speech and expressive language delay, so linking a second gene or genes to a
speech/language disorder.

In the short term, should duplication of 7q11.23 be a common cause of speech/language impairment, we would like to see genetic testing integrated into speech-language pathology and genetics clinics.

In the long term, by making the link between a speech/language disorder and a particular gene or set of genes, we will be able to learn about how those genes function in speech and language development, and how having extra copies of them can result in speech or expressive language delay. Eventually, this may help to design better therapies for such disorders, although this is a long way off.

## Where can I get more information?

For further information or for a copy of the study consent form, please contact:

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