STUDY TITLE: **The 15q13.3 research project**

INVESTIGATORS:
Radboud University Nijmegen Medical Centre:
Bregje W.M. van Bon, M.D
Bert B.A. de Vries, M.D., Ph.D
University of Washington School of Medicine:
HeatherC. Mefford, M.D., Ph.D
Evan E. Eichler, Ph.D

Introduction

Research into the causes of genetic conditions is carried out at the department of Human Genetics of the Radboud University Nijmegen Medical Centre, the Netherlands and at the Department of Genome Sciences, University of Washington School of Medicine, USA. We would like to invite you to take part in this project to study chromosome 15q13.3 microdeletions, as a member of your family is affected with this condition. We ask you to read the study information carefully and to ask any questions that you may have, before agreeing to participate.

Background information

Chromosomes are the structures in our cells that contain genes. Genes are the instructions made from DNA that allow the body to grow, develop and to maintain health. Each cell of the body contains ~25.000 genes, which are located on 46 chromosomes which come in pairs, one from each parent. They are numbered 1 to 22 from largest to smallest. The 23rd pair are the sex chromosomes, chromosome X and Y. Chromosomes are divided into two segments, the short arm (written as "p") and the long arm (written as "q"). If a part of a chromosome is missing it is called a *deletion.* 15q13.3 deletion means that there is a portion within the q arm of chromosome 15 that is missing. The deletion is called a "microdeletion", because the specific small part of chromosome 15 cannot be detected by standard testing with a microscope.

Purpose of Study

The purpose of this study is to learn more about the health problems experienced by children and adults with a 15q13.3 microdeletion, in order to be able to provide up-to-date information and the best care. You may have many questions, including questions about health outcomes and the treatment of medical problems. This study will attempt to provide better answers to these questions.

We would also like to get better insight how the 15q13.3 microdeletion results in health problems in some individuals with the deletion. Several genes in the 15q13.3 region are missing and we would like to investigate the relationship of specific health problems to these genes.

Description of the Study Procedures

If you agree to be in this study, we will ask the clinician involved in the care of you or your child to provide a medical description by filling out an information sheet. We will also ask for photographs, to define whether there are similarities in appearance between persons with this deletion.

In addition, we will ask for a blood sample from you and/or your child. Stored cells may already be available from you or your child. If so, we would ask for permission to access this

sample and additional blood samples will not be necessary. The blood or cell samples will be studied at the Department of Human Genetics of the Radboud University Nijmegen Medical Centre, Nijmegen, the Netherlands. The samples we store will not be used for any other purpose than this study. It is also possible to participate in specific parts of the study.

Risks/Discomforts of Being in the Study
The potential risks in this study are very small and may include minor discomfort while taking blood with a needle, a short period of discomfort after the procedure and a chance that there will be a bruise at the collection site.

Benefits of Being in the Study
The results of this study could be of importance for you and/or your family by providing up-to-date information and care. If there is important information for the parents or guardians, your doctor or genetic counsellor will be informed. Subsequently he/she can contact you. However, please take into account that in a research project results can take a long period of time (sometimes several years).

Costs
There are no costs to you to participate in this research study.

Confidentiality
The records of this study will be kept private. Summary study written information will be shown on the website, but individual children or adults will not be able to be identified. Photographs will not be shown on the website or published without written permission from yourself, or from the parents or guardians. Access to the records will be limited to the researchers.

In any sort of report we may publish, we will not include any information that will make it possible to identify a participant. Consent for publication will also be requested prior to publication.

Voluntary Participation/Withdrawal
Your participation is voluntary and you are free to withdraw at any time, for whatever reason. Whether you take part in this study or not, it will not make any difference to the medical care you will receive. If you do decide to take part in the study, you can still withdraw at any time and this will not make any difference to your medical care either.

Contact information
If you have further questions please contact

Bregje van Bon, M.D.
Department of Human Genetics
Radboud University Nijmegen Medical Centre
Postbox 9101
6500 HB Nijmegen
Phone: +31 24 361 3946 Fax: +31 24 361 3946
info@15q13.net

or the doctor/ genetic counsellor who initiated the research in your family.