STUDY TITLE **The 17q21.31 research project**

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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 (http://www.ru.nl/english/). We would like to invite you to take part in a project to study the 17q21.31 microdeletion syndrome, 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

The 17q21.31 microdeletion syndrome in which a small part of chromosome 17 is missing. Chromosomes are the structures in our cells that contain genes. Each cell of the body contains ~25.000 genes, which are located on 46 chromosomes. Chromosomes come in pairs one from each parent and are numbered 1 to 22. The 23rd pair contains the sex chromosomes, chromosome X and Y. The chromosomes can be 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.* A 17q21.31 deletion means that a specific part of the q arm of chromosome 17 q21.31 is missing. The deletion is called a "microdeletion", because the deletion cannot be detected by standard chromosome analysis.

Purpose of Study

The purpose of this study is to learn more about the health problems experienced by children and adults with a 17q21.31 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 and the treatment of medical problems. This study will attempt to provide better answers to these questions.

We would also like to understand how the 17q21.31 microdeletion results in health problems. Several genes in the 17q21.31 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 participate in this study, we will ask the clinician involved in the care of your child to provide a medical description of your child by filling out an information sheet. We will also ask for photographs, to define whether children and adults with the 17q21.31 microdeletion have similar features that could assist in making the diagnosis in other children in the future.
In addition, we will ask for a blood sample from your child and preferably also from both parents. Stored cells may already be available from 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 if you wish.

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 involved to participate in this study.

Confidentiality
The records of this study will be kept private. A summary of the clinical features of all children and adults with the deletion will be shown anomalously on the website of the project. Photographs will not be shown on the website or published without written permission from the parents or guardians. Access to the records will be limited to the researchers only.

In any sort of report we may publish, we will not include any information that will make it possible to identify a participant. Informed consent for publication will always 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 additional questions, please contact the doctor/ genetic counsellor who initiated the research in your family or contact:

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