Department of Women's and Children's Health
Te Tari Hauora Wāhine me te Tamariki
Clinical Genetics Research Group

INFORMATION SHEET – NON-NZ PARTICIPANT

What are the genetic causes of developmental disorders in humans?

INTRODUCTION

You/your child are invited to take part in a study on the genetic cause of developmental disorders. Your participation is entirely voluntary and if you choose not to take part this will not affect any future care or treatment of you or members of your family. If you do agree to take part you are free to withdraw from the study at any time, without having to give a reason. There will be no payment for taking part in the study.

Around 1 child in every 30 is affected by a developmental disorder. In this study we are trying to define the genetic factors that underlie some of these conditions. This involves closely examining the genetic material (DNA) of individuals to find alterations that could explain their condition. You are being asked to participate because you are affected, or are related to someone affected, by such a condition

ABOUT GENETIC STUDIES

Each person has a DNA make-up (their genes) which is different from that of everybody else. Because this research investigates genetic make-up, this information can identify a participant and their particular genetic characteristics. This information is therefore kept confidential and will not be disclosed, stored, or used in any way without the informed consent of the participant.

WHAT DOES THE STUDY INVOLVE?

Presently we are inviting individuals and their families with developmental disorders to participate in this research. Your participation in the study is entirely voluntary (your choice).

Participation involves the following:

- Having a blood sample drawn (5 ml from a child; up to 20 ml from an adult) having this genetic material extracted, stored and analysed.
- Some individuals may have a small piece of skin removed under local anaesthetic for the same purpose. These blood and/or skin cells will be grown in the laboratory so that a continuous source of genetic material is available for study and consequently no further request for blood tests would be required.

- Allowing your physician/geneticist to share medical notes with us so that we can learn about your/your child's medical history (for example clinical findings, laboratory test results and xray/scan results).
- Any subsequent use of your sample will be subject to approval from a Health & Disability Ethics Committee.
- Once such an answer is found that information will be given back to you by your doctor/geneticist. At that stage your doctor will be able to answer any questions about the results. The accumulated knowledge gained in this manner will improve our understanding of why developmental disorders occur in general.
- All genetic material and data will be stored in locked cabinets within locked laboratories. This material and information will be stored for a maximum of 30 years (or until such as time as Professor Robertson ends the study) whereon it will be either returned to you or destroyed.

BENEFITS AND SAFETY

The main benefit for participating in this study is learning what genetic alteration led to your/your child's condition and therefore a more accurate idea of inheritance pattern of the condition in your family.

RISKS

1. One risk of the study is the inconvenience and discomfort associated with a blood test and/or skin biopsy.
2. Another risk relates to the chance of making an unexpected genetic discovery regarding a participant's health status. In some situations, the genetic analysis will be focused on just a small number of genes. In other situations, a comprehensive evaluation (for example an examination of every gene – an "exome" or "genome" sequencing study) might be performed. Therefore, many potential genes may be examined to find the alteration responsible for each participant's condition. It is therefore possible we could discover something not related to the research question, but which is still relevant to that person's health or your family's health and can be acted upon by you to reduce the potential for harm. In the experience of researchers worldwide the chances of this happening are less than 1%. If this situation (called "the discovery of an incidental finding") arises we will discuss the situation with your referring clinician about the need for confirmatory testing. That clinician will then feed back this information to you and together you will decide on a course of action. Unexpected findings may vary considerably in their implications for you and your family. If the analysis reveals a risk for a condition that is not of clear and serious health importance then you will NOT be informed of such findings.

   If we do inadvertently discover something of major health significance that is reported back to you, it is important to note that this information could be considered "prior knowledge" of a medical condition. This knowledge may then potentially impact on subsequent ability to obtain life or medical insurance.

SAFETY

Participation in this study will be treated with the utmost confidentiality. All identifying information we obtain will not be disclosed without your informed consent. The researchers will not claim any right, ownership or property in your individual genetic information without your having first sought and obtained informed consent to the transfer of any such right, ownership or property. In consenting to participate in DNA sampling for the proposed study it will not be construed as creating any right or claim on the part of the researcher to your genetic information.

CONFIDENTIALITY

All samples and associated clinical details will kept in a locked filing cabinet and stored there until the end of the study whereupon it will be destroyed. No other person, other than the scientists directly involved in the study, will have access to these resources.

STATEMENT OF APPROVAL

This study has received ethical approval from the Health and Disability Ethics Committee (NZ).

Please feel free to contact the researcher if you have any questions about this study:

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