

## INFORMATION SHEET

### 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 causes of developmental disorders. Participation is entirely voluntary and if you choose not to take part this will not affect any future care or treatment. If you do agree to take part you are free to withdraw from the study at any time, without having to give a reason. If you require a whanau support person to discuss this proposal with then this can be arranged. 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/your child 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 your genetic material extracted, stored and analysed.
- On some occasions a saliva sample will be sufficient to obtain sufficient genetic material for this study.
- 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 your /your child's medical notes with us so that we can learn about your medical history (for example physical findings, laboratory test results and xray results).
- Any subsequent use of the samples obtained for this study will be subject to approval from a Health & Disability Ethics Committee.
- It is possible that the investigators may need to collaborate with overseas researchers to carry out some genetic analyses. This would involve sending a small amount of DNA overseas. In this situation the DNA samples will still be under the custodianship of the investigators and any DNA not used up in the study will be returned to New Zealand.
- You can choose a standard disposal method for the samples, or request a karakia (blessing).
- Once such an answer is found that information will be given back to you by your doctor/geneticist. At that stage this 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 of participation 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 that 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 your (or your child'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 your health, or your family's health, and can be acted upon by you in a practical way to reduce the potential for harm. In the experience of researchers worldwide the chances of an incidental finding like this being discovered are less than 1%. If this situation (called “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**

Your/your child’s 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 or that of your kinship group, *hapu* or *iwi*, 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**

The samples and all clinical details will be 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:**

### **Principal Investigator:**

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## YOUR RIGHTS

If you have any queries or concerns regarding your rights as a participant in this study you may wish to contact a Health and Disability Advocate. This is a free service provided under the Health and Disability Commissioner Act.

- Telephone: (NZ wide) : 0800 555 050
- Free Fax (NZ wide) : 0800 2787 7678 (0800 2 SUPPORT)
- Email (NZ wide) : [advocacy@hdc.org.nz](mailto:advocacy@hdc.org.nz)

## COMPENSATION

In the unlikely event of a physical injury as a result of your participation in this study, you may be covered by ACC under the Injury Prevention, Rehabilitation and Compensation Act. ACC cover is not automatic and your case will need to be assessed by ACC according to the provisions of the 2001 Injury Prevention Rehabilitation and Compensation Act. If your claim is accepted by ACC, you still might not get any compensation. This depends on a number of factors such as whether you are an earner or non-earner. ACC usually provides only partial reimbursement of costs and expenses and there may be no lump sum compensation payable. There is no cover for mental injury unless it is a result of physical injury. If you have ACC cover, generally this will affect your right to sue the investigators. If you have any questions about ACC, contact your nearest ACC office or the investigator.

## IF AN INTERPRETER IS REQUESTED

Participants will need to be reasonably fluent in English; however an interpreter may be available.

English	I wish to have an interpreter	Yes	No
Deaf	I wish to have a NZ sign language interpreter	Yes	No
Māori	E hiahia ana ahau ki tetahi kaiwhaka Māori/kaiwhaka pakeha korero	Ae	Kao
Cook Island Māori	Ka inangaro au i tetai tangata uri reo	Ae	Kare
Fijian	Au gadreva me dua e vakadewa vosa vei au	Io	Sega
Niuean	Fia manako au ke fakaaoga e taha tagata fakahokohoko kupu	E	Nakai
Sāmoan	Ou te mana’o ia i ai se fa’amatala upu	Ioe	Leai
Tokelauan	Ko au e fofou ki he tino ke fakaliliu te gagana Peletania ki na gagana o na motu o te Pahefika	Ioe	Leai
Tongan	Oku ou fiema’u ha fakatonulea	Io	Ikai