



Te Whare Wānanga o Ōtago

INFORMATION SHEET FOR NEW ZEALAND PARTICIPANT

What are the genetic causes of malformations in humans?

INTRODUCTION

You are invited to take part in a study on the genetic cause of congenital malformations. Around 1 child in every 50 is affected by a congenital malformation (birth defect). In this study we are trying to define the genetic factors that underlie some of these conditions, in particular those that lead to abnormal development of the skeleton and brain. This involves closely examining the genetic material (DNA) of individuals to find alterations that could explain their 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 identifies a participant and their particular genetic characteristics. This information is confidential and will not be disclosed, stored, or used in any way without the informed consent of the participant.

ABOUT THE STUDY

Presently doctors world wide are inviting individuals with congenital malformations to participate in this research. Participation involves having one blood sample drawn (a teaspoonful from a child; two table spoonfuls from an adult) and having the blood sent to us where genetic material will be extracted and analysed. Some of this analysis may be performed in laboratories run by our collaborators overseas. Some individuals may have a small piece of skin removed under local anaesthetic for the same purpose. It may also be decided to grow these blood and/or skin cells 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.

It is hoped that over the course of the study that many potential genes will be examined to find the alteration responsible for each participant's condition. Once such an alteration 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 malformations occur in general. All genetic material and data on you 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 RISKS AND SAFETY

The main benefit for you as a participant in this study is learning what genetic alteration led to your condition and therefore a more accurate idea of inheritance pattern of your condition in your family. The main risks of the study are the inconvenience and discomfort associated with a blood test and/or skin biopsy.

Your 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. Additional information about the study can be obtained from your referring physician or directly from the principal investigator (address below). If you require a whanau support person to discuss this proposal with then this can be arranged.

CONFIDENTIALITY

Your participation in this study will be treated with the utmost confidentiality. Your sample and all clinical details about your condition will be coded, kept in a locked filing cabinet and stored there indefinitely. No material which could personally identify you will be used in any reports on this study. 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 New Zealand MultiRegion Ethics Committee.

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

Principal Investigator:

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Please feel free to contact your local disability advocate if you have any questions or issues about this study that you may wish to discuss with an independent person:

Northland to Franklin: 0800 555 050 Mid-to-lower North Island: 0800423638
South Island (except Christchurch): 0800 377 766 Christchurch: 03 377 7501