Pūnaha Io – the New Zealand Neuro-Genetic Registry & BioBank

PARTICIPANT / PARENT INFORMATION SHEET

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This study has received ethical approval from the Health and Disability Northern A Committee (ref. 2023AM1526) and has been reviewed and approved by the Māori Research Review Committee of ADHB.

We are inviting you or your child to take part in Pūnaha Io - the New Zealand Neuro-Genetic Registry & BioBank, for people with neurogenetic and neuromuscular conditions.

Neurogenetic diseases are conditions that affect the brain, spinal cord, nerves or muscles that are caused by variants in our DNA. Neuromuscular diseases are conditions affecting the nerves and muscles and the junctions between them. While many of these are genetic some are due to inflammation or degeneration without clear cause. We use the word "Neuro-Genetic" to include both sorts of disorders.

Take your time to read the information and decide whether you wish to take part.

Introduction

The aim of this combined registry and biobank is to lower the barriers for New Zealanders with Neuro-Genetic disease to be involved in research and clinical trials. We also aim to support the establishment of best-practice care for Neuro-Genetic patients in Aotearoa-New Zealand.

Registries are databases containing clinical information about individuals who are affected by a specific condition. In rare disease, they play an important role in the therapy development pathway.

Registries can:

- Identify participants for research studies and clinical trials
- Help develop care standards, to help improve the care people receive
- Identify specific research questions
- Provide information for doctors and scientists to learn more about Neuro-Genetic diseases
- Represent a link between patients and the research community, providing the opportunity for people to receive information directly relevant to their condition

Biobanks are a useful research resource. They collect, process and store tissue samples, and body fluids for ethically approved research studies.

Major progress has been made in recent years towards finding new treatments for conditions like spinal muscular atrophy, Duchenne muscular dystrophy, Huntington's disease, myotonic dystrophy, facioscapulohumeral muscular dystrophy, limb girdle muscular dystrophies, Charcot-Marie-Tooth disease, inherited ataxias, hereditary spastic paraparesis and others Neuro-Genetic disorders.

Many of these treatments target specific genetic defects, some of which are rare and affect only relatively small numbers of people worldwide. When a trial of these treatments is being planned it is important that suitable participants can be found and contacted quickly. One way of ensuring this can happen is to collect each participant's details in a registry ahead of time. This registry contains the

important information that researchers will need in order to identify prospective study participants such as participants' genetic test results and their current functional abilities.

The data held in the registry will also help researchers answer other important questions such as how common these diseases are. It is also a way to inform participants about the latest information relevant to their disease.

By linking data held in a registry with samples donated and stored in a biobank, a powerful resource is created that can help answer questions about what happens as a participant's condition progresses over time and can also be used to identify eligible candidates to test potential therapies on.

Pūnaha Io is a patient registry and biobank that is internationally networked with the global family of TREAT-NMD patient registries, the Cure Huntington's Disease Initiative, the Rare Diseases Clinical Research Network, the Critical Path Institute, International Rare Diseases Research Consortium.

Am I eligible to participate?

All people in New Zealand, Tokelau, Niue and Cook Islands affected by Neuro-Genetic disease are eligible to enrol with Pūnaha Io. You do not have to have a confirmed genetic diagnosis but may participate:

- If your condition is recognised as being a Neuro-Genetic condition.
- If you are an adult and are related to someone with a Neuro-Genetic disease.
- If you are an adult without any neuro-genetic conditions who can act as a 'population control'

Whether you want to take part or not is entirely your choice and your decision will not affect your future healthcare. If you do agree to take part, you can change your mind at any time and we can remove your information from the database and return or destroy your samples. However, samples which have already been shared with other researchers may not be able to be retrieved.

If your affected child is able to understand what this registry is about we will also ask for their consent. When your child turns 16 we will ask for their informed consent.

What do I have to do?

If you agree to participate you will need to consider the benefits and risks involved with taking part in this study before you sign the consent form and return it to the study co-ordinator. There are some parts of the study which are required if you want to take part, and other parts that are optional. The main required part of the study is agreeing to the collection, storage and sharing of your data, and the main optional part is whether you want to give samples.

You may hold beliefs about sacred and shared values of any tissue samples removed and data originating from the tissue. The cultural issues associated with sending your tissue samples and data overseas and/or storing your tissue and data should be discussed with your family/whānau as appropriate. If you need cultural support this can be provided. Please let us know and we will arrange this for you or you can ring the number at the bottom of the participant information and consent form. Cultural support is different from knowing more about the study. However, if you do want to know more about the study we can arrange for the primary investigator to come and talk to you and your whanau.

You should consult with your family and whānau. You may wish to complete the consent form with the assistance of your doctor or community support worker or study co-ordinator. If you are not sure of the answer to any question you should discuss this with your doctor, community support worker or study co-ordinator.

The study co-ordinators will be available to discuss any other questions or concerns you have about the consent form. The study co-ordinator contact details are on page 10.

Data collection

Depending on the type of Neuro-Genetic condition being studied, data for the Registry may be collected from medical records, or through extra clinical assessments. For example, these assessments might include physical examination (coordination etc), cognitive tests, and questions about how you feel.

Sample donation

You have the option of also giving samples to Pūnaha Io. There are different types of samples that we might ask you to consider donating:

- Non-invasive biological samples, e.g. saliva, tears, urine, faeces
- Minimally invasive biological samples e.g. blood, swabs
- Invasive biological samples, e.g. cerebrospinal fluid (CSF), skin biopsy

Information regarding the risks of specific invasive procedures will be provided at that time of sampling. We will always confirm your consent at the time of sampling.

Biological samples may be obtained directly from you by appropriately qualified people. Biological samples and biopsies may also be obtained from left-over clinical samples e.g. if you have had, or are going to have a clinical muscle, skin or nerve biopsy, we will ask your permission to obtain a leftover sample of that. If you are going to have a lumbar puncture we will ask your permission to obtain some of the CSF.

We also request that you contact the study coordinator if your contact details change or there are major changes in your clinical condition.

By consenting, you agree to the study coordinators accessing your clinical record, including any genetic test results and to also contacting you to inform you about research opportunities or to update your record.

What happens to the information I provide?

The information you provide will be stored securely and confidentially in the Registry on password protected computers at Auckland City Hospital. No unauthorised people will be able to gain access to any information about you. You will be able to access, and advise of any corrections to, your own information.

Researchers will apply to access data for projects with ethical approval, and this will be overseen by the Pūnaha Io Access Committee.

Your data can be shared anonymously e.g. if a researcher wants to know how many people with your condition there are on the Registry who are over a particular age. Alternatively your data may be linked to a unique ID number so that researchers know about your individual clinical information but don't know who you are. This is called "de-identified" data. For example, if there is an international database for your condition, e.g. the CMT International Database, you will be informed of this and your de-identified data will be forwarded to that database. Researchers who access your data in such registries must have been approved by their own local ethics committee and by the relevant international oversight committee, e.g. TREAT-NMD global database oversight committee (TGDOC).

Sharing of your data with such studies where you are anonymous or de-identified is a routine function of Pūnaha Io and, therefore, will generally be decided upon by the study coordinator and principal investigator. The Pūnaha Io Access Committee will be informed of sharing with such studies.

If a clinical trial or other research study is identified for which you might be eligible, a Pūnaha Io study coordinator will contact you. If you are interested, we will share details about the trial or study with you, and you can contact the organiser of the clinical trial or study. We will never share your name or any other identifying personal details with external researchers.

You are free to make your own decision about whether or not you wish to participate in the trial, and may wish to discuss this with your doctor.

Possible risks of personal (private) information collection include the fact that, as with the collection of any personal (private) information, there is a slight risk of accidental disclosure of information or breach of computer security. Loss of confidentiality could have a negative impact on you, your family, or other individuals or groups, including affecting your insurability, employability and/or family relationships. Safeguards are in place to minimise this potential risk.

Giving biological samples

Not everyone will be asked to donate biological samples, e.g. blood. Some people may be invited to consider providing a single donation, or multiple (e.g. annual) donations.

Non-invasive sample collection

You may be invited to consider donating biological samples secreted or excreted from the body, e.g. saliva, urine, tears or faeces. The process of collecting such bio samples is not invasive and therefore of very low physical risk.The process will be fully explained at the time of collection.

Minimally invasive sample collection

You may be invited to consider donating samples that can be easily collected by minimally invasive techniques, e.g. blood, cheek/nasal swab. Blood samples are obtained in the same way that you might have a regular blood test. With your consent we would arrange for a blood draw of approximately two teaspoons (10mls) of blood from you. Your body replaces the small amount of lost blood within 24 hours.

Possible risks associated with blood collection include experiencing pain and/or bruising at the site where blood is taken. A clot may form at the site and infections may occur, but these are rare. Fainting or feeling lightheaded may occur during or shortly after having blood drawn. If you experience this, you should lie down immediately to avoid possible injuries and notify study personnel.

Invasive sample collection

Depending on the Neuro-Genetic condition that you have you may be invited to consider donating samples collected by an invasive procedure, e.g. skin biopsy, muscle biopsy or cerebrospinal fluid (CSF). If you are considering these types of sample donation, you will be provided with an additional information sheet which details the sampling process and the risks involved. All invasive biosampling will be carried out by a trained clinician under sterile conditions and using local anaesthetic.

In brief, a skin biopsy takes a small piece of skin 5 mm in diameter in a procedure that takes about 10 minutes.

A muscle biopsy requires a small cut in the skin, exposing the muscle and then a piece of muscle is cut out, about the size of a matchstick head. The skin is then closed up with some stitches. This procedure takes about 10-20 minutes.

CSF is the fluid that bathes your brain and spinal column. CSF can be used to provide information about the brain and the nervous system that is impossible to obtain in any other way. CSF is collected by a procedure called a lumbar puncture or spinal tap. This is a commonly performed procedure that takes around 30 minutes. Up to 10mls of CSF is withdrawn. Your body replaces the lost CSF within 24 hours.

We will always confirm your consent at the time of sampling and collection will only be undertaken with your consent.

What happens to the biological samples I provide?

Prior to each sample collection the study coordinators will provide you with full information regarding the sample collection procedure and what will be stored in the biobank, and obtain consent from you to collect and store your sample to be used for research in the future. You can choose each time whether you want to donate a sample or not.

The study coordinators will arrange for the collection of the sample. Generally, the sample will then be sent to Te Ira Kāwai the Auckland Regional Biobank, where trained staff will prepare the samples for storage. The sample will be labelled with a unique laboratory code which will be linked to the corresponding information about you in a separate password-protected table.

If you have donated samples that contain cells (e.g. blood, urine, tissue biopsies), these cells may be isolated, grown and stored in a way that allows them to be further grown as ‘primary cells’ or ‘cell lines’, e.g. skin biopsies can be used to isolate skin cells (primary fibroblasts) which can be grown and studied; or induced pluripotent stem cells (iPSCs) can be generated from these skin cells.

The samples will be stored until there is a request for the samples to be used. If the sample is to be used in a way where you will not be identified, then the Te Ira

Kāwai Scientific Advisory Board will receive a recommendation from Pūnaha Io Access Committee and then make the final decision whether to release the samples to the requesting researcher.

Occasionally external studies will require samples which need to be processed immediately before they are biobanked e.g. when there is something in the blood that won't survive freezing and storage. In such cases the sample will not go to Te Ira Kāwai and the responsibility for the samples will remain with Pūnaha Io. We will only be doing this if the Access Committee has agreed in advance that the samples can be shared with the researcher. You will be informed if this affects your samples.

You can also choose whether your sample remains in New Zealand or can be sent to researchers overseas as well.

Typical uses for your samples would be to look for markers of disease progression, or to help develop new treatments. Genetic testing may be carried out on the sample. If these genetic tests are carried out in a research lab the results will not usually be available to participants.

If the sample and/or your information is to be used in a way where you may be or will be identified, the study coordinator will contact you. You will be free to make your own decision about whether you wish to have your samples and/or information used in this way.

You are under no obligation to provide any of these types of samples at any time. You can change your mind about donating samples at any time. If you change your mind after having donated a sample, we are able to withdraw your samples from the Biobank destroy them with or without karakia (prayer).

What happens if I don't want to be part of the study anymore?

In the event that you do withdraw from this study, no new information about you will be collected or added to the study database; however, the information you have already provided will continue to be stored, used, and shared in the manner described in this form, **unless you request that this information be deleted**. Such information will not be deleted if (1) it is not possible to identify you from this

information, or (2) if your de-identified information is needed to preserve the integrity of this study or for reasons of public interest in the area of scientific research. In the event that you do withdraw from this study, the biological samples collected from you during this study will continue to be stored, used, and shared in the manner described in this consent form, **unless you request that the samples be removed from the storage facility and destroyed**. If any of the biological samples collected from you during this study have already been distributed for use, **it will not be possible destroy them.**

Whom do I contact for further information?

Thank you for making the time to read about and consider taking part in Pūnaha Io – the New Zealand Neuro-Genetic Registry & BioBank. We hope you will be interested in enrolling.

For more information, please contact the study coordinators by
Email: neurogenetics@adhb.govt.nz
Phone: 021896662
Post: **Pūnaha Io - New Zealand Neuro-Genetic Registry & BioBank**
Neurology, Auckland City Hospital,
Private Bag 92024,
Auckland Mail Centre 1142.

If you have any queries or concerns regarding your rights as a participant in Pūnaha Io, please contact the Study Coordinator or the Principal Investigator, as above.

If you want to talk to someone who isn't involved with the study, you can contact an independent health and disability advocate:
Phone: 0800 555 050
Email: advocacy@advocacy.org.nz

You can also contact the health and disability ethics committee (HDEC) that approved this study on:
Phone: 0800 438 4427
Email: hdecs@health.govt.nz

If you require Māori cultural support contact the administrator for He Kamaka Waiora (Māori Health Team):
Phone: 09 307 4949 ext 29200.
State the title of the study and name of the primary investigator.