N.Z. Cardiac Inherited Disease Registry

Information for Individuals / Families Considering Enrolling

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Introduction

You are invited to take part in the Cardiac Inherited Disease Registry. Participation in the Cardiac Inherited Disease Registry is voluntary, that means it is your choice whether or not you wish to be part of it. Individuals and families will be offered the opportunity to be included in the Registry when they are referred to Specialists around New Zealand.

Please take the time to read through this information sheet and specific information sheet about the Cardiac Inherited Disease affecting you or your family. If you have any questions feel free to contact the Registry co-ordinator, or ask your Specialist.

Background

Some heart diseases can be inherited; passed down through generations in a family from parents to children, at times appearing to skip a generation. But genes cannot skip generations, they are simply passed on or not (most of the time for every child born in a family where there is a cardiac inherited disease present; there is a 50% chance of inheriting a gene or not), however sometimes people can inherit the genes but never be affected by the condition and this is called being a carrier. Conversely others in the family will inherited the gene for the condition and will become symptomatic with it. Carriers and those affected by a condition are both able to pass the condition on to their children. This is why even those who do not have any symptoms should undergo clinical evaluation since you may potentially carry a condition that you do not know that you have.

The Cardiac Inherited Disease Registry is being created in response to the need for a specialist co-ordinated service for affected families and the clinicians that look after them. We hope to make the process of identifying and screening potentially affected family members easier and more efficient for the families and doctors alike.

Cardiac inherited diseases vary in severity, even within a family. Some individuals may develop only a very mild form of the disease, while others in the same family may become very ill, or even die unexpectedly, sometimes at a young age. We believe that families with Cardiac Inherited diseases will be better managed if they are included in a disease or condition-specific registry.

The Registries for each of the conditions will be co-ordinated by Jackie Crawford, who is The Cardiac Inherited Disease co-ordinator in New Zealand, based at Auckland Hospital. Her work will be medically supervised by Associate Professor Jon Skinner, Paediatric Electrophysiologist (Children's Heart Rhythm Specialist)/Cardiologist, at Starship Hospital in Auckland.

Which Cardiac Inherited diseases will be included in the Registry?

Currently we have included the following conditions, Long QT syndrome, Hypertrophic (HCM) Cardiomyopathy and Dilated Cardiomyopathy (DCM), Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT), Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC), Brugada Syndrome, and Sudden Unexplained Death in the Young (SUDY) "Looking for answers."

Other Cardiac Inherited diseases, which may be added in the future include: Familial Premature Coronary Artery Disease, Familial Dyslipidaemia, and Marfans Syndrome. Each Cardiac Inherited disease will have its own specific information sheet.

What is the Purpose of setting up a Registry?

The purpose of setting up this Registry is to collect information to help patients and families affected by Cardiac Inherited Disease, and the Doctors, who are looking after them. Specifically the aims are to:

- Co-ordinate standardised diagnostic screening tests for relatives of affected individuals.
- Provide a reliable source of information to individuals, families and the medical community about specific cardiac inherited diseases.
- Offer genetic (DNA) testing when and where this is available. This may enable early diagnosis to be made that may enable appropriate treatment and prevention strategies to be undertaken.
- Initiate gene-specific therapies if they become available.
- Assess the prevalence of Cardiac Inherited disease, to assist our knowledge and assist with healthcare planning and resource provision.

FREQUENTLY ASKED QUESTIONS ABOUT CARDIAC INHERITED DISEASE REGISTRY:

1) What benefits are there in joining the Cardiac Inherited Disease Registry?

Belonging to the specific Cardiac Inherited Disease registry may have several benefits for you and your family.

- Firstly you and or your Doctors will be advised of information/medical advances which are relevant to your specific Cardiac Inherited Disease.
- We will undertake regular screening of people on the Registry, and keep you informed about new tests and treatments being formulated for your specific condition.
- The Registry will store your blood sample locally, with your consent, and this sample will be used for diagnosis/research into your specific cardiac inherited disease. If and when genetic analyses new diagnostic tests or treatments become available we will be able to contact you as soon as possible.
- The Registry will update its records regularly, by staying in contact with you.
- The registry will provide access to validated information including information on support groups and information web sites, to assist you and your family, keep track of new knowledge specific to your condition.

1) What do I have to do to join the registry?

Usually we prefer you to be referred by the Specialist who is looking after you or your family; however some General Practitioners may refer patients. The Specialist can either contact the Registry directly, or send your information to either the co-ordinator or to Dr Skinner. You will be given information sheets at the clinic, or these will be posted to you. You are entitled to have any questions you may have, answered and to take as much as you need to decide whether or not you wish to participate.

If you do decide to join the Registry, you will need to sign a formal consent form to indicate that you are participating voluntarily, and that you understand the purpose and the process of the Registry you are joining.

2) Can I withdraw from the Registry if I want to?

Yes, participation in the Cardiac Inherited Disease Registry is completely voluntary (your choice) you can decide to cease participation at any time, by notifying us in writing.

3) Can children join the Registry?

Yes, we encourage children to join the registry. We believe it is good to include the child in the decision to join the Registry, especially if a Cardiac Inherited disease affects the child. It is important that parents choose a place to do this where they feel the child will be receptive and comfortable; often this will not be in the hospital setting.

Children from the age of 8 up to 16 may be able to consent to joining the Registry and therefore sign their own name on the registry consent form; however, we still require parents or guardians to sign it as well. When a child reaches the age of 16 years, we are required to obtain consent from them, to continue in the registry as an adult.

Each member of a family will need to sign a consent form if they wish to participate in the Registry. In the case of small children or those rendered incapable of giving informed consent for any reason, we rely on the parents or guardians to make the decision in the best interests of the person on their behalf.

4) What happens if I choose not to enrol in the Cardiac Inherited Diseases Registry?

If you choose not to be involved in the registry, you will continue to be seen by your usual Specialist Physician. Your medical condition will be managed in the usual manner.

If one family member chooses to cease on-going participation in the registry, the other family members will still remain enrolled unless they too send us notification that they wish to cease participation.

5) Who will have access to my information?

Only the Registry co-ordinator and the Doctors directly involved in your case will have access to your information.

Your information will be coded and stored on a specific Registry Database. The coded information will be password protected and unable to be accessed by any other users. We have taken every precaution to ensure that your information remains private.

No information that specifically identifies you or any members of your family will be given to anyone other than your referring Doctors. Any information that is published by the registry will not identify individuals or families by name or any other identifying feature.

Should you ask us to release information to other Doctors of your choice we will do this for you but can only accept written and signed requests for release of information?

6) Are there any risks involved, in joining the Cardiac Inherited Disease Registry?

There is no physical risk involved in joining a data registry. However you should consider the possible consequences of joining a long-term data registry. You will be contacted from time to time to provide updated information, in order to keep the records current. Being aware that you may carry a Cardiac Inherited Disease may cause you to feel many mixed emotions. We believe the best way to deal with these is to keep informed about your condition; however some people may not wish to be made aware of new information.

You need to consider whether you wish to know, if you do carry genes that cause a Cardiac Inherited disease.

You should also be aware at the outset that results may take months or many years to become available. We have no way of predicting when researchers will find the answers we are waiting for. However the registry will be able to provide you with information as new findings come to light.

Some genetic testing is able to be performed in New Zealand, however depending upon the condition and the analyses that need to be performed the tests may have to be done outside of New Zealand, where large research facilities specialise in specific tests or techniques. You will be informed if your particular condition requires testing to be performed overseas. If testing is required to be performed overseas, the samples will be rendered anonymous by substituting a letter/number code in place of your name and other details. Keeping samples anonymous is a way of protecting your information, and as already stated we undertake to keep all your information private.

In the unlikely event of a physical injury as a result of your participation in the Registry you will be covered by the accident compensation legislation with its limitations. If you have any questions about ACC please feel free to ask the researcher for more information before you agree to take part in the Registry.

7) What are the implications of knowing you have a Cardiac Inherited Disease?

There are aspects to life with such a diagnosis, which makes some people choose not to know the results of screening tests (such as Electrocardiograph (ECG), or Echocardiogram or a genetic diagnosis (the blood test results). They may put their own lives at risk by choosing not to know however at least their results are helpful in the context of the whole family.

If you are diagnosed with a Cardiac Inherited Disease you may experience problems associated with obtaining Insurance. In general Insurance companies like to be made aware if you have any health problems, which may run in your family, as soon as you become aware of them. If you have any specific concerns regarding insurance you should discuss your questions directly with your Specialist.

There may be some other aspects of receiving a diagnosis that may cause you to have some limitations placed on your lifestyle. Limitations could include: activities where it was felt that you may be placing yourself at significant risk, such as competitive sporting activities, or driving or operation of heavy industrial equipment.

Family members, who are being treated for a Cardiac Inherited Disease, should follow the guidance they receive from their Specialist in this regard. If you have any questions about restrictions that you may be asked to comply with if you are diagnosed with a specific Cardiac Inherited Disease, you should discuss them directly with your Specialist.

If you do decide to join the Registry you will be asked to decide whether or not you wish to know the results of your gene tests if and when a diagnosis is made.

8) Are there any risks associated with tests to screen for my Inherited Cardiac Disease?

If you have not previously had any screening tests arranged to check for the presence of an inherited cardiac disease, your Doctor will probably want to arrange some. There is usually no difference between the tests your Doctor will arrange and the tests we arrange on his/her behalf, if they have not already been done.

If you do choose to join the cardiac inherited disease registry, we will ask you for a blood sample, so that we may extract a DNA sample. The blood will be used for the specific purpose of obtaining a genetic diagnosis of your cardiac inherited disease, either by already available, or tests which are being developed by research laboratories.

There are minor associated risks to having blood tests; such as pain, stress, infection and blood loss, however major complications from having a blood test would be very rare. For those who experience discomfort during blood testing a cream called Emlar can be applied around the site the blood will be drawn from approximately an hour before the blood test is done, to minimise discomfort.

There are variable degrees of risk associated with the current diagnostic screening tests. The risk is different for each procedure, but ranges from the risk of skin abrasion (for an Electrocardiograph ECG) through to the risk of death approximately 1: 100,000 for Patients' undergoing Exercise Tolerance Tests (ETT). If you have any specific concerns about having a test done, you should discuss it with your Specialist prior to having the test.

The most common tests used include Electrocardiograph (ECG), Echocardiogram (Ultrasound viewing of the heart) Holter Monitoring (24-48 hours of continuous ECG data), and Exercise Tolerance Testing (ETT) which monitors the ECG as you exercise. Most commonly patients may only need one or two tests in order to screen for the specific disease, however your Specialist will decide which tests you should have.

9) Are there any costs associated with joining this Registry?

There is no cost to you in joining the Cardiac Inherited disease registry. If you wish to post information you will be required to pay the cost. If you wish to make a telephone enquiry, please leave your name and contact details on our answering machine we will ring you back.

10) How long will the Cardiac Inherited Diseases Registry operate for?

The Cardiac Inherited Disease registry will operate as long as there remains a need for it to do so, and as long as funding remains available.

If you are interested in joining the Cardiac Inherited Disease Registry, you should also read the **specific information sheet**, outlining the features of the Cardiac Inherited disease you or your family is suspected of having. If you have any questions relating to your decision to participate in the Registry please feel free to either contact the Registry co-ordinator and/or discuss them with your Specialist.

If you have any queries or concerns regarding your rights as a participant in the Cardiac Inherited disease registry you can contact an independent Health and Disability Advocate. This is a free service provided under the Health & Disability Act; through the offices of the Health and Disability Commissioner.

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

The N.Z Cardiac Inherited Disease Registry has received Ethical Approval from the N.Z. Multi-centre Ethics Committee: AKX/02/00/107