Sudden death from a genetic heart disorder

Approximately 100 sudden natural deaths occur in 1–40 year olds in New Zealand each year. In about half of these, either no cause is found ('unascertained') in the post-mortem examination or there may be signs of a heart muscle problem ('cardiomyopathy').

Could somebody else in my family die of the same thing?

Most disorders are not genetic. In these cases, other family members are safe and can be reassured.

In about one-third of the unascertained cases, there's an underlying genetic problem. In these cases, other family members may be at risk.

When a heart muscle problem is found, up to half of family members will be expected to carry the same gene. Once doctors identify people at risk, they can be helped.

Heart muscle problems

If a heart muscle problem is found, the pathologist may suspect that the death is related to a sudden catastrophic heart rhythm disturbance caused by a genetic disorder in the heart that no one knew about. The pathologist and the coroner may ask for help from the Cardiac Inherited Disease Group (CIDG), a national team of heart and gene specialists.

The CIDG investigation

If the CIDG investigates, they will send family members a letter, inviting them to attend a clinic to meet the heart and gene specialists. If possible, this will be done locally so the family doesn't have to travel. It is helpful if a number of family members can come at the same time.

The specialists will talk about the post-mortem findings, take a family history, and ask for any further information about the deceased person's medical history. Some simple heart tests, such as an ECG (electrocardiogram) and heart scan (echocardiogram), will be offered to close relatives, such as parents, siblings and children.

There may be medical tests (blood, tissue and genetic) on samples saved from the post-mortem examination. The results of these tests are reviewed by the specialist team, including the pathologist, and passed onto the family and coroner.

HOW LONG DOES IT TAKE?

The pathologist will usually refer a case to the CIDG more than 3 months after a death. They have to wait for special laboratory test results from the post-mortem examination and then write a report.

The coroner will usually release the death certificate and post-mortem report to the family before the CIDG investigation, so the family can make all practical and financial family arrangements.

The CIDG investigation can take many months. Genetic tests take about 2–3 months, and organising the clinics and all the cardiac tests sometimes takes longer, depending on the health service resources and how widespread the family is.

DO FAMILY MEMBERS HAVE TO TAKE PART?

No. This is a clinical service designed to help people find a cause of death for their family member, and screen them to see if they are also at risk. Attendance is strongly recommended, but there is no legal requirement.

The CIDG coordinators are happy to talk on the phone first. Because different family members may be in different stages of grief, or may not want to attend with certain other family members, the CIDG will try to work with the family's needs.

What will the investigation achieve?

If the death was caused by a genetic condition, the CIDG investigation will find if other family members are affected. Death can then be prevented through daily medication or, in rare cases, a special pacemaker-defibrillator implant.

What sort of cases does the CIDG investigate?

The most common genetic conditions causing sudden death without leaving any signs are long QT syndrome, Brugada syndrome and 'CPVT'. Heart muscle conditions include hypertrophic cardiomyopathy (HCM), dilated cardiomyopathy (DCM) and arrhythmogenic right ventricular cardiomyopathy (ARVC).

The CIDG investigates:

- sudden unexplained death in otherwise healthy young people with no significant post-mortem findings
- seizure-related deaths with unusual features
- sudden unexplained death in good swimmers or when someone saw the deceased collapse in the water
- post-mortem findings of possible or probable inherited cardiomyopathy (HCM, DCM, ARVC)
- sudden deaths associated with medications, supplements and drugs that can prolong the QT interval.

DOES THE CIDG INVESTIGATE SUDDEN INFANT DEATHS (SIDS)?

Not usually. Extensive research in New Zealand has shown that fewer than 10% of SIDS cases are related to conditions like long QT syndrome, and it is extremely uncommon for other family members to be at risk.

This means the CIDG is only asked to investigate if the case is unusual, like if the infant died while awake or none of the known risk factors (such as co-sleeping) were present.

The pathologist will usually recommend the long-term storage of DNA, in case more information is found later on and the family wants it to be investigated.

How can I get more information?

Families are usually referred to the CIDG by a pathologist or coroner, but you can contact the CIDG at any time. Talk to the coronial case coordinator for more information.

Once you've been referred to the CIDG, you can contact your local CIDG coordinator or ask the CIDG administrator for advice.

YOUR LOCAL CIDG COORDINATOR

South Island

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CIDG ADMINISTRATOR

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ABOUT THE CIDG

The service is led by Associate Professor Jon Skinner, Childrens Heart and Rhythm Specialist, Starship Children's Hospital. The heart specialists work closely with the national clinical genetic service.

Details of the clinical teams and further advice about the heart conditions can be found on www.cidg.org or through a coronial coordinator.