Family Heart Screening by a Cardiologist

Family heart screening aims to find heart problems which may run in the family, and which may have caused the sudden death in the young family member. Heart screening involves meeting a heart doctor (cardiologist) and undergoing some tests to look at the structure, electrical rhythm and function of your heart.

Why is family heart screening important?

If no cause for the sudden death was found at autopsy, family screening tests may reveal that another family member has a condition which could not only explain the death of the relative, but also be putting him or her also at risk of sudden death. Effective treatments are available to prevent sudden death.

If a familial ("inherited") heart condition was diagnosed at autopsy, then the tests on the family will be chosen to look for that condition.

Due to the potential risk to family members, we advise all first-degree relatives (children, parents, siblings) to be assessed by a cardiologist with expertise in inherited heart diseases.

A visit to the cardiologist must include a clinical history being taken, physical examination, and some special tests such as those described below.

What happens at family heart screening?

Firstly the cardiologist will interview the family, or some family representatives. He or she will explain why this screening is recommended, and will inform you what he/she knows of the autopsy report results. This will be also your chance to ask questions. The cardiologist will want to know more details about the events surrounding the death of your relative, and will ask for details of any previous medical conditions or treatments. These are important because there may provide clues as to the diagnosis. For example frequent blackouts associated with swimming might point to long QT syndrome or a condition called CPVT. You may remember some medical test results done during life that could be obtained from another hospital and reviewed- an ECG (electrocardiogram) for example- your permission to obtain these may be required.

The doctor or a colleague (nurse/counsellor etc) will take a family medical history and draw up a family tree with your help - a history of premature deaths, fainting or of conditions such as epilepsy for example may be very important. Someone in the remote family with a history of recurrent blackouts may be at risk and need to be reviewed in case they are at risk.

What types of tests may be involved?

All of the tests are voluntary; you do not have to have these tests. However, none of them are painful or harmful, but they do a take a little time to do and then to report, and it may not be possible to do them all at the same clinic visit.

Electrocardiogram (ECG)
An ECG is a recording of the heart rhythm that involves placing ECG electrodes (stickers) on the arms, legs and across the chest. A recording is then made showing the electrical activity of the heart. The test takes 3-5 minutes and requires you to be lie still.

Echocardiogram (Echo)
An echocardiogram is an ultrasound of the heart that allows the cardiologist to look at the structure, size and contraction, valves and flow of blood through the heart. This takes about 30-40 minutes.

Exercise test
This test assesses how the heart responds to exercise (i.e. when your heart is required to work harder than normal). This requires you have an ECG while walking and running on a treadmill. Blood pressure is monitored during and after exercise and also sometimes the amount of oxygen in the blood. In some cases an echocardiogram will be done before and after exercise. This test takes around 30-40 minutes.

Holter Monitor
A Holter monitor is a small mobile ECG recorder which allows you to wear it for long periods of time (24-48 hours), and examines the electrical activity of your heart throughout the day. The recorder is about the size of a Walkman and has ECG electrodes (stickers) which attach to the chest. The recorder must not get wet, so a bath or shower is not possible until it is removed, however you are otherwise able to get on with your daily activities. You will be asked to keep a diary of activities while wearing the monitor, it will take around 15 minutes to fit the monitor and explain the procedure.

Cardiac Magnetic Resonance Imaging (cMRI)
A cMRI uses a magnetic field and radio waves to give a very detailed picture of the heart. It is done in a special facility, so a separate appointment would need to be made for this test. It allows cardiologists to evaluate parts of the heart that may not be assessed as adequately with other technology. You will need to remove all jewellery and other metal objects for the test. The cMRI takes about 45 minutes.

What if nothing shows up in the family heart screening?

Unfortunately we are unable to give a definite answer as to the cause of death in more than half of the families we see, because we find no clear evidence of a familial heart problem. Sometimes special genetic tests can be helpful, either done on some of the blood or tissue from the deceased, or from a blood test on a family member (see…link) These will be discussed with you.

If after all the tests nothing is found, then it's possible that the cause of death was due to a heart or other problem that does not run in families. However, since we cannot be sure, and since some heart conditions become worse later in life, continued regular check-ups are usually offered to first-degree family members every few years. Sometimes the level of suspicion of a familial heart problem is relatively low and it may be considered safe to not have further follow-ups.