Familial Dilated Cardiomyopathy (FDC)

In dilated cardiomyopathy, the left ventricle (the main pumping chamber of the heart) becomes enlarged and the muscle of its wall is thin. This affects its ability to pump blood effectively (shown below). Heart failure can occur if the problem is severe and causes symptoms such as shortness of breath, swelling of the ankles and legs, and fatigue. Sometimes dilated cardiomyopathy will affect the electrical system of the heart and lead to rhythm problems, which may require implantation of a pacemaker. Rarely, dilated cardiomyopathy will cause sudden death.

Dilated cardiomyopathy has many causes, such as coronary artery disease, viruses, alcohol and thyroid disease. A cause is found in 50% or more of those affected but not in the remainder. Familial dilated cardiomyopathy (FDC), the situation in which there is more than one affected person in a family, accounts for 20-50% of undiagnosed cases. FDC is caused by inherited gene alterations and makes investigation of the family very important.

The severity of symptoms can vary greatly within a family (i.e. one family member may suffer severe heart failure or even sudden death, while another may be only mildly affected and have minimal problems). For this reason, it is recommended that all first-degree relatives (parents, siblings, children) of someone with FDC are assessed by a cardiologist, who may recommend further checks at regular intervals. Early detection is important, so that the condition can be monitored and if necessary, treated to try to prevent future complications.

How it is inherited
FDC is usually inherited as an autosomal dominant condition but can sometimes be inherited as an X-linked condition (with males more likely to be affected). See section on Genetic Inheritance for more information.

Genetic testing
Genetic testing for FDC is not straightforward because of the number of genes (over 30) that have been associated with the disorder. Testing is available but is expensive and is
not yet routine. Improvements in technology, including development of new cardiac gene chips will hopefully make testing more readily available.

**Treatment**

Treatment in dilated cardiomyopathy aims to improve heart function. Medications can be very effective. In severe cases, a special cardiac pacemaker treatment known as “resynchronisation” can be very helpful. A small group of people with FDC who are considered at high risk of cardiac arrest may benefit from an ICD (implantable defibrillator). Whether these treatments are appropriate for you should be discussed with your Cardiologist.

It is recommended that people with FDC should see their cardiologist at least once a year.

**For more information**

Further information is available from the Cardiomyopathy Association of Australia at www.cmaa.org.au

You can also find out more about genetic heart conditions and the latest research news at the Australian Genetic Heart Disease Registry at www.heartregistry.org.au or the Cardiac Inherited Diseases Group (New Zealand) at www.cidg.org