THE MATER HOSPITAL

The Inherited Cardiac Conditions Clinic at the Mater Misericordiae University Hospital

Irish Inherited Cardiac Conditions Network

Mater Family Heart Screening Clinic

The Mater Family Heart Screening Clinic has been set up to cater for families affected by unexplained Sudden Cardiac Death in a young person primarily to establish the cause of death. We also assess other family members for the presence of the same condition and protect them as needed with medications or an implantable cardiac defibrillator. The clinic also assesses patients who may have or have been diagnosed with or have a family history of an inherited cardiac condition.

Inherited Cardiac Conditions

These include inherited or genetic conditions that affect the structure of the heart muscle called Cardiomyopathies or the function of the electrical channels within the membrane surrounding each heart muscle cell known as Channelopathies. Cardiomyopathies may cause an individual to have chest tightness, shortness of breath, palpitations, fainting spells or rarely cardiac arrest which may be fatal. Patients with Channelopathy may have fainting spells or rarely cardiac arrest but usually don't have the other symptoms. Because they are genetic, inherited cardiac conditions can be passed from a parent to a son or daughter.

Another group of conditions that affect the main blood vessel coming from the heart, the aorta with aneurysm (swelling) or dissection (a tear in the wall) referred to as Aortopathies can also be inherited and run in families.

Other conditions that cause structural abnormalities evident from birth referred to as congenital heart disease (e.g. Atrial septal defect, Teralogy of Fallot) may or may not be inherited.

Dyslipidaemias are a group of genetic conditions that increase the cholesterol level and make people prone to developing arterial disease which can affect the coronary arteries and cause a myocardial infarct (a heart attack). Lastly, there are a number of inherited conditions that affect the heart as well as other organs such as the liver, kidneys or the nervous system (e.g., Muscular Dystrophies, Glycogen storage diseases, Fabry's Disease).

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Cardiomyopathy

There are 3 main cardiomyopathies.

In **Hypertrophic Cardiomyopathy (HCM),** the muscular wall of the left ventricle thickens to > 15mm and may become fibrosed or scarred. This can reduce the pumping function of the heart and can make the heart electrically unstable. The condition may be suspected on the basis of any of the symptoms above and an abnormal ECG but requires imaging with an echocardiogram or cardiac MRI scan to make the diagnosis.

Dilated Cardiomyopathy (DCM) causes the heart's left ventricle to stretch in size with reduced contractility / pumping function and eventually a reduced cardiac output. It typically causes shortness of breath but can cause palpitations, fainting or cardiac arrest. The diagnosis is made on echocardiography or cardiac MRI scan

Arrhythmogenic Cardiomyopathy causes fibrosis or scarring of the right or left ventricle and sometimes dilatation of just the right ventricle. It usually presents with arrhythmias that cause palpitations, fainting or less commonly cardiac arrest. The diagnosis may be suggested by ECG or echocardiography but usually requires a cardiac MRI scan.
Exercise can provoke dangerous arrhythmias in patients with cardiomyopathy so strenuous exercise should be avoided. Treatment is usually with beta blockers, antiarrhythmic drugs or an implantable cardiac defibrillator. Other medications to improve heart function and breathing such as ACE inhibitors and diuretics can also be used.

Channelopathy

There are a number of Channelopathies all of which are diagnosed of ECG changes either at rest, with exercise or with intravenous drugs such as adrenaline for LQTS and CPVT or Ajmaline for Brugada syndrome. The commonest Channelopathies are;

Long QT Syndrome (LQTS) which is diagnosed when the QT interval which can be measured on a standard ECG is greater than 480ms. It can also be diagnosed with less prolonged QT intervals if other factors such as a family history of LQTS or recurrent fainting spells are present. The condition can cause exercise or stress / emotion-related fainting spells or even cardiac arrest usually in the first 20 years of life although it may present later. Treatment is usually with beta blocker medications, avoidance of QT prolonging medications (see 'www.crediblemeds.org' for medications to avoid in LQTS), and Potassium supplementation and in high risk patients an implantable cardiac defibrillator (ICD).

Brugada Syndrome causes a typical ECG abnormality which may be provoked with infusion of a drug, Ajmaline. This is done as a day case test in the hospital with careful monitoring. Fainting episodes occur at rest or cardiac arrest if it occurs can happen during sleep. Some medications can cause the ECG changes. The list of medications to be avoided is available on the website

‘www.brugadadrugs.org’. Treatment of Brugada syndrome in patients at high risk is usually with an ICD.

Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) is similar to LQTS but without the ECG abnormality. The diagnosis is usually made with the development of ventricular arrhythmias on exercise.

Less common channelopathies include **Short QT syndrome** (similar to LQTS but treatable with different medications) and **J wave or Early repolarisation** syndromes (similar to Brugada syndrome but with their own distinct ECG pattern).
All of these conditions may present with an unexpected fainting spell usually with no warning or with a cardiac arrest which can be triggered by exercise or emotional upset or by certain commonly prescribed medications. Vomiting / diarrhoea and Potassium loss can also trigger arrhythmias in all 3 conditions as can fever in Brugada Syndrome.
Beta blockers can prevent arrhythmias dramatically in LQTS, Quinidine can be helpful in Brugada, J wave and early repolarisation syndromes and Flecainide can reduce arrhythmia burden in CPVT.

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