Cardiac Conditions - Cardiomyopathy

A cardiomyopathy is defined as a disorder in which the heart muscle is structurally and functionally abnormal, in the absence of coronary artery disease, high blood pressure, valve disease and congenital heart disease sufficient to cause the observed abnormality. Cardiomyopathies can be familial/genetic, occurring in more than one family member, or non-familial/non-genetic, occurring in the patient and not affecting other family members. Types of cardiomyopathy include hypertrophic cardiomyopathy, dilated cardiomyopathy, arrhythmogenic right ventricular cardiomyopathy, restrictive cardiomyopathy and unclassified cardiomyopathies.

Hypertrophic cardiomyopathy (HCM) is perhaps best known for being the most common cause of sudden cardiac death in young athletes, but it also affects roughly 1 in 500 of the general population and is associated with sudden cardiac death in seemingly fit and healthy adults. There are a number of gene mutations responsible for HCM and genetic testing can now be used to identify family members at risk of inheriting the disorder. Many affected families have an autosomal dominant pattern of inheritance, meaning that there is a 50% chance of any first degree relative (sibling, parent, child) carrying the affected gene. The way the abnormal gene “expresses” itself in any one patient, however, is highly variable; some patients may have no symptoms at all and heart function will appear normal, whereas others may be very limited at an early age by symptoms such as breathlessness, palpitations, chest pain, fatigue and fluid retention.

If a patient is found to have hypertrophic cardiomyopathy, it is important that the rest of the family is screened so that appropriate advice and treatment can be offered, including drug therapy, pacemakers and implantable cardioverter-defibrillators (ICDs) in selected patients, and techniques to remove the thickened heart muscle by conventional surgery (myectomy) or percutaneously (septal ablation).

Dilated cardiomyopathy (DCM) is not uncommon; at least 25% of patients in Western populations have evidence for familial disease with predominantly autosomal dominant inheritance. Diseases linked to the X sex chromosome, such as Becker and Duchenne muscular dystrophies, are also associated with DCM, as are other gene mutations. There are also many non-familial causes such as infection of the heart muscle (myocarditis), caused by a virus, excessive and prolonged alcohol ingestion or nutritional deficiencies. Muscle recovery may occur in patients whose cardiomyopathy has a specific and reversible cause, such as alcohol-induced and virally-mediated cardiomyopathy.

Restrictive cardiomyopathy has many causes and is characterised by increased stiffness of the heart muscle, typically affecting the filling of the heart chambers with blood. Arrhythmogenic right ventricular cardiomyopathy is a very specific abnormality which, although uncommon (1 in 5000 prevalence), is a frequent cause of sudden death in young people in some parts of Europe.

All disorders of heart muscle form and function need careful evaluation to determine the precise abnormality, the value of genetic testing and screening of family members, and to direct appropriate treatment to alleviate symptoms and prevent future complications.