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THE ROLE OF DEFIBRILLATORS IN CHANNELOPATHIES

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The channelopathies, a group of disorders that can cause sudden death, may affect children and adults. Patients may be identified when investigated for loss of consciousness or by screening. These disorders cause ventricular tachycardia and ventricular fibrillation which in many cases can be aborted by shocking. Implantable defibrillators may be life-saving but may also be responsible for complications that can significantly impact on quality of life. Decisions are challenging as some patients are at higher risk than others. Defining who will benefit from a defibrillator is difficult and the stakes are high.

When considering whether to implant a defibrillator in a young patient it is important to weigh up the risks and benefits for that patient as an individual. Children and younger patients have a greater number of problems with defibrillators than their older counterparts due to lead fracture, inappropriate shocks and implant/device related complications. Inappropriate shocks can be extremely difficult for the patient.

The channelopathies have been called the "silent assassins" as they can present in an otherwise healthy person with a sudden and unexpected lethal arrhythmia, leaving no trace at subsequent autopsy. These disorders may be inherited or arise de novo in the patient and therefore screening of family members of affected patients plays an important part of management. The best known channelopathies are long QT syndrome, Brugada syndrome and catecholaminergic polymorphic ventricular tachycardia.

Perhaps the best known channelopathy is long QT syndrome, estimated to affect 1:2000 people. Twelve different types have been described based on the genes identified to be causal. Most genes recognised code for proteins involved in the function or architectural make-up of ion channels responsible for ion movement within the myocardial cell. The different genetic types differ in their clinical course, morphology on ECG, trigger for arrhythmia and response to therapy.

Types 1, 2 and 3 account for 90% of genotyped Long QT syndrome and have been most studied. With the help of international registries established, we now have a greater understanding of many aspects of the condition. Risk stratification is important to enable us to identify patients who are not protected by beta blocker therapy alone and who might need an implantable defibrillator. Age and gender play a part in determining risk but perhaps the most important is QT length itself. High risk groups include: male children, adult women with Long QT 2 and patients with a QTc > 500. It would seem that Long QT 3 patients derive less protection from beta blockers. Recent research in Long QT 2 has shown that the position of the mistake in the protein, i.e. the individual mutation location, can give us further guidance in terms of risk and response to therapy. Those patients who have inherited or are born with two mutations are at risk, often present in infancy.

Peter Schwartz published the results of the European Long QT registry patients implanted with a defibrillator in 2010. 91% of recipients had been symptomatic with approximately half being survivors of cardiac arrest. However 9% were asymptomatic many of whom were long QT 3. 28% of patients received an appropriate shock, but in 11% they were inappropriate.

Brugada et al. described another channelopathy which is thought to account for the greatest number of sudden adult deaths. The only proven effective therapy is an implantable defibrillator and so those considered to be at high risk will benefit. Those at low risk may have a two and a half times greater risk of having an inappropriate than an appropriate shock in addition to other defibrillator complications. Risk stratification in Brugada syndrome is less clear than in long QT. Those survivors of a prior VF arrest would appear to be at high risk and should therefore have a defibrillator. Those who have had syncope or ventricular tachycardia also appear to be at high enough risk to make implantation of a defibrillator beneficial. Those asymptomatic patients either with a type 1 ECG at rest or revealed on administration of a drug are difficult to assess and the risk-benefit ratio of defibrillator implantation is controversial. Children may be affected by Brugada syndrome and although in general perceived to be at lower risk than adults can still be at risk. Decisions for this group are even more difficult.

Catecholaminergic polymorphic ventricular tachycardia (CPVT) is more likely to present in childhood than adult life with exercise related syncope or sudden death. Early recognition and intervention is very important to save lives. With a normal resting ECG the condition may be mistaken as epilepsy. Beta blockers are the treatment of first choice and many derive good protection, but not 100%. For those refractory to beta blocker therapy or those unable to tolerate beta blocker, left cardiac sympathectomy and/or a defibrillator needs to be considered. However, defibrillators are less than ideal therapy for this condition. The rhythms encountered are more diverse in CPVT and are difficult for current defibrillators to recognise correctly and accurately, leading to a high risk of inappropriate shocks. Arrhythmia may be self terminating so a defibrillator should be programmed to allow for this rather than

shocking early, this goes some way to limiting unnecessary shocks. Moreover arrhythmias seen in CPVT are often triggered as opposed to re-entry in mechanism so may not always respond to shocking.

Once the decision to implant is made, the type of defibrillator needs to be chosen. A conventional defibrillator with intracardiac lead can be employed from about 7 years upwards. In a small child an epicardial system needs to be considered. Because of the problems with intracardiac leads, the subcutaneous defibrillator has been developed and can be considered in children >30kg. With every device careful programming is an absolute requirement to attempt to limit inappropriate shocks.

In conclusion, the decision to implant a defibrillator needs to be made after careful definition of the individual patient's own risk. Choice of device is important. Strategies to reduce device related risks should be employed. Careful programming is imperative. Other therapies should be used alongside defibrillator therapy to limit the number of shocks required. With great care defibrillators can be used in affected children to save lives.

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