Genetic profile of inherited arrhythmias in Hong Kong children

Dr. Tak-cheung YUNG
Consultant Paediatric Cardiologist & Electrophysiologist
Chief of Service, Department of Paediatric Cardiology, Queen Mary Hospital
Division of Paediatric Cardiology, Department of Paediatrics and Adolescent Medicine,
The University of Hong Kong, Hong Kong

Long QT syndrome, short QT syndrome, Brugada syndrome, catecholerminergic polymorphic ventricular tachycardia are now increasingly recognized in paediatric age group. They are arrhythmic problems with clear genetic causes due to mutations of a single gene. On the whole, they are uncommon but very important causes of sudden cardiac death. Patients with these clinical diagnoses should have investigation of the genetic profile. The genetic diagnosis can confirm the clinical suspicion. When the index case is confirmed, predictive testing for relatives will be able to identify other mutation carriers within the family. With medical treatment and preventive measures, symptoms and sudden death in the at-risk relatives may be prevented. Within each genetic arrhythmia syndrome there are numerous subtypes according to the genetic mutation. The risk of sudden death of each subtype is related to the specific gene, as well as to the mutation type and location. Genetic diagnosis also facilitates cardiologists to determine the best treatment option for patients with inherited arrhythmias. For example, clinical studies have demonstrated that different beta-blockers have different efficacy in reducing symptoms in each long QT syndrome subtypes.

In this presentation, the development of genetic testing and genetic profile of inherited arrhythmias in Hong Kong children will be discussed.