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INHERITED ARRHYTHMOGENIC CHANNELOPATHIES AND IMPLANTABLE CARDIOVERTER DEFIBRILLATOR TREATMENT

National and age-related perspectives

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Akademisk avhandling

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Title
Inherited arrhythmogenic channelopathies and implantable cardioverter defibrillator treatment: national and age-related perspectives.

Abstract
Background Long QT syndrome (LQTS) and Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) are two hereditary diseases caused by mutations in genes coding for cardiac ion channels. Patients may be at risk of symptoms such as syncope and sudden cardiac death (SCD). To prevent symptoms, high-risk patients may receive an implantable cardioverter defibrillator (ICD). ICD treatment prevents SCD, but it also carries a risk of ICD harm: inappropriate shocks and complications. In these studies, we investigated ICD treatment in patients with inherited arrhythmogenic channelopathies with a focus on risk assessment and ICD harm. **Methods** Studies I–III use registries to identify patients and medical records to collect data. Studies I and III focus on phenotype, genotype, and medical treatment (Study I) and the diagnostic process and treatment over time (Study III). In Study II, we assess risk scores and ICD harm. Study IV is a systematic review using a data synthesis without meta-analysis. **Results** In Studies I and II, most patients received ICD treatment after serious events according to guidelines, although, asymptomatic patients also had ICD treatment from 2006. We could not validate available risk scores and a high percentage of patients were affected by ICD harm. In Study III, many CPVT patients had been misdiagnosed at first with improvements over time. The treatment of CPVT patients also changed over time in accordance with guidelines. In Study IV, 20% of all children had inappropriate shocks and 24% had other complications. However, in LQTS and CPVT patients, the percentage of patients with ICD harm was lower in studies published from 2015, than before. **Conclusion** These studies illustrate both the difficulty in pre-ICD risk stratification and its important role due to high combined rates of ICD harm. We found improvements in how patients have been managed over time and identified the need for future research addressing benefit and harm from ICD treatment among children and adolescents.

Keywords
Long QT syndrome, catecholaminergic polymorphic ventricular tachycardia, risk score, implantable cardioverter defibrillator, appropriate shock, inappropriate shock, complication, children, adolescents, systematic review, Sweden.

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