



### **G19 Genetic Aspects of Sudden Death in Youth: A Retrospective Study of Familial Hypercholesterolemia**

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After attending this presentation, attendees will understand some principles of genetic heart disease and the advantage of genetic examination in selected forensic autopsies of sudden death. Preliminary results of premature ischemic heart disease will be presented as an example.

This presentation will impact the forensic science community by serving as a key aspect of sudden cardiac death investigation as it can augment traditional means of investigation by including postmortem genetic examination in order to reveal familial hypercholesterolemia (FH) in young people dying from coronary athero-thrombotic disease.

Several cases of sudden death due to basis of genetic heart disease have inspired this newly started retrospective study. The goal of the study is to examine inherited heart disease from selected forensic autopsies.

Purified DNA from blood of approximately 230 selected autopsies; aged 0-40 will be examined. The following genetic heart diseases will be emphasized: Ischemic heart disease due to FH caused by defects in the low density lipoprotein receptor (LDLR) and apo – lipoprotein B (ApoB) gene; Long QT-syndrome and Brugada syndrome due to defects in cardiac ion channel proteins; catecholaminergic polymorph ventricular tachycardia due to defects in the ryanodine receptor; arrhythmogenic right ventricular cardiomyopathy due to defects in the desmosome proteins; hypertrophic, and dilated and restrictive cardiomyopathies due to defects in the contractile proteins.

Preliminary results of the study concerning premature ischemic heart disease will be presented. Examination of approximately forty cases of death in youth due to ischemic heart disease is being examined for defects in the LDLR and ApoB gene.

Mutations in the genes of the above mentioned proteins are known to present as arrhythmia or sudden death. Diagnosed cases of sudden cardiac death in the Danish population are few, despite the estimated higher number of cases in the literature. The perspective of the study is to determine the molecular cause of sudden cardiac death in order to intervene and prevent sudden cardiac death in relatives to cases with proven genetic heart disease.

**Sudden Cardiac Death in Youth, Genetic Heart Disease, Familial Hypercholesterolemia**