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Natural and Undetermined Sudden Death: Value of Post-mortem Genetic Investigation

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Background. Sudden unexplained death may be the first manifestation of an unknown inherited cardiac disease. Current genetic technologies may enable the unraveling of an etiology and the identification of relatives at risk.
Objective. The aim of our study was to define the etiology of natural deaths, younger than 50 years of age, and to investigate whether genetic defects associated with cardiac diseases could provide a potential etiology for the unexplained cases.
Methods. Our cohort included a total of 789 consecutive cases (77.19% males) <50 years old (average 38.6±12.2 years old) who died suddenly from non-violent causes. A comprehensive autopsy was performed according to current forensic guidelines. During autopsy a cause of death was identified in most cases (81.1%), mainly due to cardiac alterations (56.87%). In unexplained cases, genetic analysis of the main genes associated with sudden cardiac death was performed using Next Generation Sequencing technology. Genetic analysis was performed in suspected inherited diseases (cardiomyopathy) and in unexplained death, with identification of potentially pathogenic variants in nearly 50% and 40% of samples, respectively.
Conclusions. Cardiac disease is the most important cause of sudden death, especially after the age of 40. Close to 10% of cases may remain unexplained after a complete autopsy investigation. Molecular autopsy may provide an explanation for a significant part of these unexplained cases. Identification of genetic variations enables genetic counseling and undertaking of preventive measures in relatives at risk.