Abstract: P2830

Clinical and genetic findings in relatives to young sudden cardiac death victims without post-mortem examination (autopsy)

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Background: Guidelines recommend clinical assessment of relatives to young sudden cardiac death (SCD) victims (<50 years) to protect the surviving relatives by pre-symptomatic interventions, in case the SCD was due to an inherited cardiac disorder. The etiology is an inherited cardiac disease in about 50% of young SCD cases. The work-up of relatives is generally guided by findings in the SCD victim. If post-mortem examinations (autopsies) have not been performed the work-up of relatives is challenged. The diagnostic hit-ratio of screening of relatives under these circumstances is unclear.

Purpose: To assess the diagnostic yield of inherited cardiac diseases of cardiac work-up in relatives of SCD victims, where no autopsy had been performed.

Methods: This retrospective study consecutively included families referred to our tertiary referral centre, specialised in hereditary cardiac diseases, during the period 2005 to 2018 due to SCD in the family. No autopsy had been performed in any of the SCD victims. The relatives underwent standard cardiac work-up according to guidelines. Based on the findings in the relatives the families were categorised into: 1) definite diagnosis, 2) borderline diagnosis or 3) undiagnosed.

Results: We assessed 149 relatives (43±16 age, 48% men) to 84 SCD un-autopsied cases (44±11 age, 79% men). In 11 (13%) families a definite diagnosis was established, in 8 (10%) families a borderline diagnosis was found and the remaining 65 (77%) families remained undiagnosed. The most common diagnosis was premature IHD (36%) followed by cardiomyopathies (27%) and channelopathies (27%). A disease-causing mutation was identified in 3 families out of 15 genetically examined families.

Conclusion: Systematic cardiac work-up of relatives to not-autopsied SCD victims, revealed a definite hereditary cardiac disease in 13% of the referred families, and a borderline diagnosis in additionally 10% of the families. Despite a reduced diagnostic yield in family members of non-autopsied SCD victims, work-up of relatives is clearly still justified.