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Comprehensive cardiogenetic testing for families of sudden unexplained death victims can save lives, scientists say

Vienna, Austria: Relatives of a young person who dies suddenly should always be referred for cardiological and genetic examination in order to identify if they too are at risk of sudden death, a scientist told the annual conference of the European Society of Human Genetics today (Tuesday 26 May). Dr. Christian van der Werf, a research fellow at the Department of Cardiogenetics, Academic Medical Centre, Amsterdam, The Netherlands said that, although his team's research showed that inherited heart disease was present in over 30% of the families of sudden unexplained death (SUD) victims, the majority of such relatives were currently not being referred for examination.

When an individual aged 1-50 years dies suddenly, autopsy reveals an inheritable heart disease in the majority of the victims. But in approximately 20% autopsy does not reveal the cause of death. "We thought that cardiological and genetic examination of surviving first degree relatives of these SUD patients might reveal an inherited heart disease", said Dr. van der Werf.

In the largest such study to date, the team looked at the outcome of first degree relative screening in 127 families who had suffered an SUD and where either there had been no autopsy (53.8%), or the autopsy did not reveal a cause of death. The average age at death of the SUD victims was only 29.8 years old.

The initial examination of the relatives consisted of taking personal and family medical history and a resting ECG. A second cardiac autopsy of the SUD victim was undertaken if tissue had been stored and was available. Additional cardiological examinations of the relatives were performed where necessary. Genetic analysis of the associated candidate gene(s) was performed in material obtained from the deceased person or in those relatives who showed clinical abnormalities.

The researchers found inherited heart disease in 36, or 32% of the families. These results meant that doctors were able to treat affected relatives and try to prevent their succumbing to sudden cardiac death. "The scale of heart disease that we found in such families underlines the necessity for general practitioners to refer first degree relatives of SUD victims to a specialised cardiogenetics department as soon as possible", said Dr. van der Werf. "Currently we estimate that only 10% of SUD families are being examined for inherited heart conditions.

The study is the second report from the registry of families who attended the Amsterdam centre's cardiogenetics department because of unexplained sudden death of a relative aged 1-50 years. The scientists intend to continue to report the yield of family screening in an increasing number of families.

"At present we are conducting a study to stimulate general practitioners and other involved physicians to request autopsy and DNA-storage for SUD patients and to refer relatives to a cardiogenetics department after a case of sudden death at young age. We hope this will lead to identification of more families at risk of sudden cardiac death, in which preventive measures then can be taken" said Dr. van der Werf.

"Relatives of young sudden death victims are often referred to cardiologists for cardiological examination. We believe relatives should instead be referred to cardiogenetics departments, where clinical geneticists, cardiologists and psychosocial workers cooperate. These professionals specialise in inherited heart diseases and their clinical and psychosocial implications, and can provide a better quality of care. Additionally, cardiologists should receive more education in inherited heart diseases. By taking these measures we can save lives and avoid further distress for families who have already suffered enough," he said.

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