



Postmortem genetic testing uncovers causes of SCD in young individuals

Sudden cardiac death (SCD) among children and young adults is an uncommon, but tragic, event. A new study published in *The New England Journal of Medicine* now reports that autopsy investigation combined with genetic testing and family screening substantially increases the identification of the cause of SCD in children

and young adults compared with autopsy examination alone.

A prospective, population-based, clinical study of SCD among individuals aged 1–35 years in Australia and New Zealand was performed to determine the causes of death after an autopsy investigation. Cases of unexplained cardiac death — defined as SCD for which no cause was found after histological and toxicological autopsy examinations — underwent additional genetic analysis to detect variants in ≥ 59 cardiac genes.

Of the 490 cases of SCD identified, 72% involved boys or young men. On autopsy, a structurally normal heart (that is, no cause of death identified, or unexplained cardiac death) was observed in 40% (198) of cases, coronary artery disease in 24%, inherited cardiomyopathies in 16%, myocarditis in 7%, and aortic dissection in 4%. Unexplained cardiac death was more likely to be observed in females and younger individuals, and occurred mostly during sleep or night-time hours. A pathogenic or probably pathogenic cardiac gene

variant was found in 31 (27%) of the 113 cases of unexplained cardiac death in which genetic analysis was performed. During follow-up, clinical screening was performed in 91 of the 198 families in which an unexplained SCD had occurred. A definite clinical diagnosis of inherited cardiovascular disease was identified in 12 (13%) of these families.

“Future research should try to explain why males are more likely to have SCD, why sleep or rest is a trigger for SCD events, and what are the causes of the remaining sudden unexplained death cases in the young,” proposes Chris Semsarian, lead investigator of the study. “The ultimate goal is to prevent SCD at all ages, and especially the young”.

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ORIGINAL ARTICLE Bagnall, R. D. et al.

A prospective study of sudden cardiac death among children and young adults. *N. Engl. J. Med.* **374**, 2441–2452 (2016)

FURTHER READING Schwartz, P.J. & Dagradi, F. Opinion: Management of survivors of cardiac arrest — the importance of genetic investigation. *Nat. Rev. Cardiol.* <http://dx.doi.org/10.1038/nrcardio.2016.104> (2016)