

The Molecular Autopsy - Beneficial in Sudden Cardiac Death

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Rates of autopsies have declined significantly in most hospitals in the US and abroad, but lessons still can be learned, especially in the era of molecular medicine.

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Source: Bagnall RD, Weintraub RG, Ingles J, et al. A prospective study of sudden cardiac death among children and young adults. *N Engl J Med.* 2016;374:2441-2452; doi:10.1056/NEJMoa1510687. See [AAP Grand Rounds commentary by Dr. David Spar](#) (subscription required).

This study describes efforts of investigators at several institutions in Australia and New Zealand to identify the underlying cause of sudden cardiac death (SCD) in individuals 1 - 35 years of age with the aid of traditional autopsy, genetic testing for known heritable causes of SCD, and investigation of surviving family members. The core of the data comes from 198 individuals classified as unexplained (no identifiable etiology after traditional autopsy) SCD, of whom 113 underwent genetic analysis. Of those, 31 had a clinically relevant cardiac gene mutation found. Furthermore, in investigation of family members of 98 of the SCD cases, 12 families were found to have members with an inherited cardiovascular disorder, including arrhythmogenic diseases and cardiomyopathies.

Autopsies seem to be destined for extinction, with rates below 1% of hospital deaths in most countries that report such statistics. It's hard to know whether that is good or bad; compared to the bad old days, imaging and other studies during life are much improved and likely lessen the benefit of autopsy in finding an unexpected important diagnosis nowadays. Still, [one study](#) of autopsies resulting from deaths occurring in pediatric and neonatal intensive care units reported a 20% rate of unexpected diagnoses gained from autopsy, including a level 1 (potentially life-threatening) cause in 5%.

Studies like Bagnall, et al should remind us that selected conditions, such as SCD, have important ramifications for surviving family members. Primary care providers are a trusted resource for families. PCPs faced with a grieving family in the setting of unexplained SCD should be proactive in recommending autopsy with genetic testing. It might just save lives.

Further Reading

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