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"Molecular autopsy" from formalin-fixed tissue

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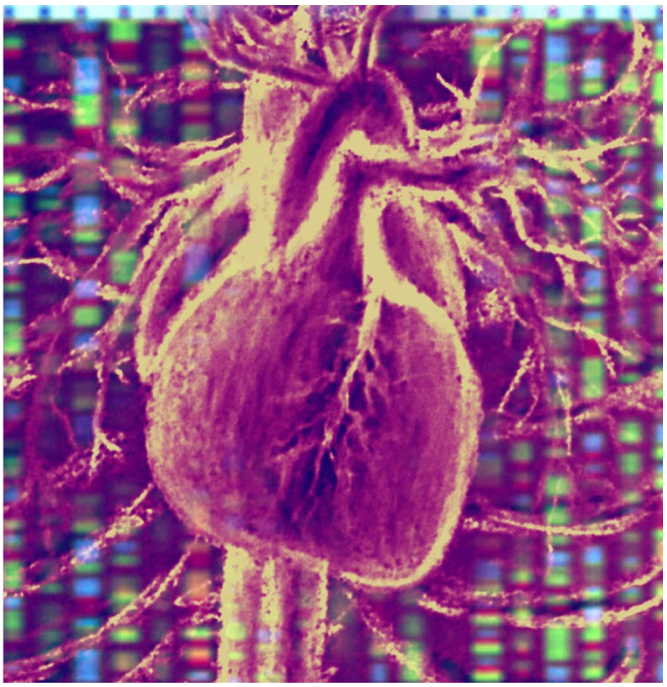

Rachel Howard

A research team in Australia reports that formalin-fixed tissue can yield DNA of sufficient quality to allow a "molecular autopsy" in unexplained cases of sudden death. The study shows that postmortem tissue can serve as a source of DNA for exome sequencing–based studies. The researchers recruited five families referred to the Genetic Heart Diseases Clinic at Royal Prince Alfred Hospital, Sydney, following the sudden death of a family member. In each case, fixed postmortem tissue blocks were the only available source of DNA. Exome sequencing recovered high-quality DNA in all five cases, despite the tissue having been stored from 3 to 14 years, and revealed the likely underlying genetic defect in three of the five cases. In two of those cases, exome sequencing confirmed previous clinical diagnoses and affected clinical management of surviving family members. The study demonstrated that DNA extracted from fixed postmortem tissue can be exome-enriched and sequenced using a commercial sequencer. However, the DNA extracted from fixed tumor tissue required additional sequencing to achieve adequate coverage of target regions equivalent in quality to that obtained with higher-quality DNA. Although whole blood is preferred for DNA extraction and genetic analysis, this study provides a precedent for turning to fixed postmortem tissue for exome-based genetic analysis when blood is not available. —*Karyn Hede, News Editor*

The ACMG reinforces its commitment to evidence-based guidance

Currently, *GIM* is the only journal with a focus on genomics and health services. As such, it is well-positioned to disseminate findings from high-quality health services research that addresses genomic medicine interventions, including outcomes research, comparative effectiveness research (CER) and systematic evidence reviews (SERs) that address genomic medicine interventions. This type of research is critical to informing the high-quality genetic and genomic medicine practice. The ACMG is committed to issuing evidence-based guidance for genomic medicine interventions, and to achieve this goal two types of documents will be produced by ACMG committees and published in *GIM*:

- clinical or laboratory practice guidelines informed by a systematic literature review (SER)
- clinical or laboratory practice resources (CPRs) informed by a non-systematic literature review

The editors of *GIM* have developed an author checklist adapted from the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) statement to assist authors in their reporting of SERs and CPRs and to help with critical appraisal by reviewers and editors. We look forward to receiving manuscripts describing health services and implementation research of genomic medicine interventions. —*Maren T. Scheuner, Editor, Genetics in Medicine*

Rachel Howard