

Postmortem testing options for individuals suspected to have suffered sudden cardiac death due to cardiomyopathy or arrhythmia

OPTION 1:

Exome sequencing

Whole Exome Sequencing (WES) is performed to detect variants in protein-coding DNA. The exome will be sequenced to an average read depth of >120x. Over 97% of the exome will be fully covered at ≥20X read depth.

How to order: submit clinical test requisition form (SLH test code 895M64) to laboratory with specimen.

Specimen requirements: 2-10 ml whole blood collected in EDTA (purple-top) tubes.

Additional information: CPT code 81415; Turn-around-time 4-6 weeks. Call laboratory for pricing.

OPTION 2:

Next Generation Post Mortem (NextGenPM) program

The Next Gen PM program is a gratis service, created to provide genomic analysis of individuals with sudden cardiac death. This program requires consent from the family prior to testing and aims to make potential results available to at risk family members for targeted variant testing.

How to participate: Immediate blood relatives of decedent should contact the Inherited Arrhythmias Clinic at UW Hospital for an appointment. This recommendation may be included in the autopsy report.

- ◆ Adult Inherited Arrhythmias Clinic at the Heart and Vascular Care Clinic scheduling number: 608-263-1530
- ◆ Pediatric Inherited Arrhythmias Clinic scheduling number: 608-263-6420

At the time of appointment the family, if interested, can be consented for testing of the decedent, if appropriate, by the cardiac genetic counselor (Kate Orland, MS CGC).

Specimen requirements: 2-10 ml Whole Blood collected in EDTA (purple-top) tubes.

Other ideal tissues sources (in additional to whole blood submission): Heart (minimum 2mmx2mmx2mm) fresh tissue into cytogenetics transfer media or minimum cell culture media (MEM) and/or Skin (deep skin punch minimum 2mmx2mmx2mm) fresh tissue into cytogenetics transfer media

Additional information: CPT code N/A; Turn-around-time 4-6 weeks

At risk family members will have the option to be tested for identified pathogenic/likely pathogenic variants using Targeted Variant Analysis (TVAR). This test is clinically available through the UW Collaborative Genomics Core (SLH test code 893M53).

Questions can be addressed to:

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