
Postmortem Genetic Testing for Sudden, Unexplained Death

Reference Guide for Medical Examiners

How can postmortem genetic testing help?

Determine cause of death: For cases of unexplained death, genetic testing may provide a cause of death up to 35% of the time.

Protect surviving family members: If genetic testing identified the cause of death, at-risk family members may undergo genetic testing to determine if they inherited the same genetic risk factor. They can receive medical care to reduce their risk.

Sample for DNA Extraction

Please obtain a **blood sample in purple top sodium/EDTA tube**. We encourage obtaining this sample if there is a possibility you may pursue genetic testing. If unavailable, we may be able to attempt DNA extraction from alternative samples. Please see backside of page for further details.

Indications

Our team is happy to provide input on any cases you are considering for genetic testing. Please consider postmortem genetic testing especially in these cases:

- Decedents age 40 years old and younger
 - Sudden unexplained death, especially during exercise or sleep
 - Cause of death possibly obscuring a cardiac etiology (i.e. drowning, single motor vehicle accident, or unexplained seizure)
 - Decedents at any age
 - Cardiomyopathies (hypertrophic, idiopathic dilated, restrictive, arrhythmic)
 - Thoracic aortic aneurysm or dissection
 - Suspected genetic condition
 - Sudden cardiac death with family history of sudden death or inherited heart disease
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Uniformed Services University MiCAP Postmortem Genetics Team

Genetic testing may be available at no cost for decedents with a history of active duty military service or who were eligible to be seen at a military treatment facility at the time of their death (servicemembers and beneficiaries). We coordinate postmortem genetic testing, result interpretation, discussion with family, and subsequent clinical recommendations. Please contact us to verify eligibility and with questions.

Contact Us: MiCAP.genetics@usuhs.edu | (301) 312-2467

Postmortem Genetic Testing Sample Storage & Shipping

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<p>Ideal Sample: 1-3mL blood in sodium/EDTA (purple top) tube</p>	<p>Short Term (<4 weeks after autopsy):</p> <ul style="list-style-type: none">• Store in refrigerator (4°C)• Ship at room temperature (in hot weather, include ice pack) <p>Long Term (months to years)</p> <ul style="list-style-type: none">• Store in freezer (-20°C to -70°C)• Ship overnight frozen in styrofoam container packed tightly with dry ice <p><i>Please ship samples on weekdays (Monday-Thursday).</i></p>
<p>Alternative Sample Types</p>	<p>If submitting backup sample type, please contact us to discuss details and limitations.</p> <p>In decreasing order of preference:</p> <ul style="list-style-type: none">• Sodium Fluoride tubes• Sodium Heparin tubes• All other tubes• Red top tubes (serum tubes)• Tissue<ul style="list-style-type: none">◦ Heart and muscle preferred◦ If <24 hours, skin biopsy preferred◦ Liver and spleen NOT preferred (limits testing options)• Paraffin Embedded Tissue• Bloodstain card/Extracted DNA?

Reference Guidelines

Deignan, J. L., De Castro, M., Horner, V. L., Johnston, T., Macaya, D., Maleszewski, J. J., ... & ACMG Laboratory Quality Assurance Committee. (2023). Points to consider in the practice of postmortem genetic testing: A statement of the American College of Medical Genetics and Genomics (ACMG). *Genetics in Medicine*, 25(5), 100017.

Middleton, O., Baxter, S., Demo, E., Honeywell, C., Jentzen, J., Miller, F., ... & MacLeod, H. (2013). National Association of Medical Examiners position paper: retaining postmortem samples for genetic testing. *Academic Forensic Pathology*, 3(2), 191-194.

Stiles, M. K., Wilde, A. A., Abrams, D. J., Ackerman, M. J., Albert, C. M., Behr, E. R., ... & Wang, D. W. (2021). 2020 APHRS/HRS expert consensus statement on the investigation of decedents with sudden unexplained death and patients with sudden cardiac arrest, and of their families. *Heart Rhythm*, 18(1), e1-e50.