

When should samples be collected for genetic testing?

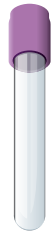

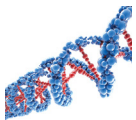

According to the Heart Rhythm Society/European Heart Rhythm Associated Expert Consensus Statement on the State of Genetic Testing for the Channelopathies and Cardiomyopathies, genetic testing is recommended for sudden unexplained death or sudden infant death where a cardiomyopathy or channelopathy is suspected. Genetic testing may be considered for any autopsy-negative case.

Why should I collect the samples?

- Providing a molecular confirmation of a clinical diagnosis allows other family members, who may also be at risk for sudden cardiac death, to be assessed for their deceased family member's disease-causing genetic mutation.
- Up to 50% of sudden cardiac death (SCD) victims under 35 years of age have no warning signs or family history of SCD.¹
- Recent studies demonstrate that genetic testing helps to identify inherited cardiac disease in 25-35% of cases of SCD in the young.^{1,2,3}
- 15% of sudden infant death cases have been shown to have an underlying genetic cause.⁴

What sample should I save?

These directions pertain specifically to cardiology genetic testing and are meant to guide you towards the most appropriate specimen for testing. If you need information about specimens for a different testing area or have questions about a specific sample, please call GeneDx at (301) 519-2100 and speak to a genetic counselor.

Sample type	Specimen details	Storage instructions	Shipping instructions	Special instructions
 <p>Blood</p>	<p>8-10cc (mL) of fresh blood in EDTA tubes. Obtaining useable DNA is not guaranteed.</p>	<p>Can be refrigerated up to 7 days before shipping. Blood specimens older than 7 days can still be shipped. GeneDx will extract DNA and test if the yield and quality permit molecular testing.</p>	<p>Ship as soon as possible at ambient temperature for overnight delivery with arrival Monday-Saturday. In hot weather a cool pack can be enclosed.</p>	<p>Blood specimens will be rejected if hemolyzed, clotted, or drawn in a non-preferred tube. If blood has been frozen, it must be shipped on dry ice to ensure that it arrives frozen.*</p>
 <p>Tissue</p>	<p>>50mg of flash-frozen tissue - preferably heart, liver, kidney, lung, or muscle. Obtaining useable DNA is not guaranteed.</p>	<p>Flash-freeze and store at -80 degree C.</p>	<p>Ship frozen on dry ice for overnight arrival on Monday-Saturday. Specimens must arrive frozen.*</p>	<p>Please call first. Fixed (paraffin embedded) tissue will be rejected.</p>
 <p>DNA</p>	<p>Minimum of 20µg (50 ng/µL) of good quality DNA.</p>	<p>Ship DNA in sealed and protective packaging at ambient temperature, using a cool pack in hot weather.</p>	<p>An express courier service is recommended for tracking purposes.</p>	<p>DNA concentration and volume MUST be provided on the specimen tube. DNA extracted from fixed tissue will be rejected.</p>
 <p>Cultured cells</p>	<p>2 T25 flasks Obtaining useable DNA is not guaranteed.</p>	<p>Cultures must be topped off with sterile medium immediately before shipping. Always establish and keep backup cultures.</p>	<p>Ship overnight at ambient temperature, to arrive Monday-Saturday. In hot weather a cool pack can be enclosed.</p>	<p>Do not send backup cultures unless requested to do so by GeneDx.</p>

*For more information on obtaining dry ice please contact GeneDx.

Where do I send the sample?

Please call GeneDx ahead of time to discuss the samples collected and review the best protocol for sending the samples. These samples are very precious and we want to be sure that all of the appropriate steps are taken during this process to preserve the integrity of the sample. After contacting GeneDx, please ship the sample overnight to:

GeneDx, Inc
207 Perry Parkway
Gaithersburg, MD 20877
(301) 519-2100

Shipping costs are generally the responsibility of the sender. GeneDx does not accept responsibility for shipping costs associated with samples sent to us.

To order a GeneDx shipping kit, please go to [genedx.com/supplies](https://www.genedx.com/supplies)

Who pays for this test?

Insurance companies do not typically cover postmortem testing and GeneDx cannot bill insurance in these cases. For pricing inquiries, please contact zebras@genedx.com. For questions about billing or payment plans, please call GeneDx at (888) 729-1206, option 2 and ask to speak with the billing department regarding payment for postmortem testing.

How can I store DNA for later use?

DNA remaining at GeneDx after completion of testing may be available for DNA banking. DNA banking is not a service provided by GeneDx, however, sending DNA to a clinical DNA bank may be arranged. Please contact GeneDx at (888) 729-1206 and ask to speak with a cardiology genetic counselor for details, or visit the Genetic Testing Registry website for more information on DNA Banking: <https://www.ncbi.nlm.nih.gov/gtr/>

What additional resources are available for families?

GeneDx recommends genetic counseling for families that pursue postmortem cardiac genetic testing. Local genetic counseling resources can be identified at www.nsgc.org. For more details, contact GeneDx at (888) 729-1206 and ask to speak with a cardiology genetic counselor.

References:

1. Tester DJ, et al. The molecular autopsy: Should the evaluation continue after the funeral? *Pediatr Cardiol*. 2012 Mar 33(3):461-70. Epub 2012 Feb 4.
2. Behr 2003 et al "Cardiological assessment of first-degree relatives in sudden arrhythmic death syndrome." *Lancet*. 2003 Nov 1, 362(9394):1457-9.
3. Tan, et al. (2003). "Sudden unexplained death: Heritability and diagnostic yield of cardiological and genetic examination in surviving relatives." *Circulation*. 2005 Jul 12;112(2):207-13. Epub 2005 Jul 5.
4. Ackerman MJ, Priori SG, Willems S, Berul C, Brugada R, Calkins H, Camm AJ, Ellinor PT, Gollob M, Hamilton R, Hershberger RE, Judge DP, Le Marec H, McKenna WJ, Schulze-Bahr E, Semsarian C, Towbin JA, Watkins H, Wilde A, Wolpert C, and Zipes DP. Heart Rhythm. 2011 Aug, 8(8):1308-39. HRS/EHRA Expert Consensus Statement on the State of Genetic Testing for the Channelopathies and Cardiomyopathies. This document was developed as a partnership between the Heart Rhythm Society (HRS) and the European Heart Rhythm Association (EHRA).