# Cardiovascular Diseases Genetic Testing Program

## Cardiovascular Diseases Single Gene Sequencing Analysis

The Molecular Genetics Laboratory at Cincinnati Children's Hospital is pleased to introduce Cardiovascular Diseases Single Gene Sequencing Analysis. This test is designed for the physician who 1) wants to order the sequencing analysis for a single gene which is part of a next-generation sequencing panel from the Cardiovascular Diseases Genetic Testing Program or 2) wants to order the sequencing analysis for any other gene which is associated with a cardiovascular related disorder.

Genes offered but not limited to:

*A2ML1, ABCA3, ABCC9, ACTC1, ACTN2, ACVR2B, ACVRL1, AKAP9, ANK2, ANKRD1, BAG3, BCL9L, BMPR2, BRAF, CACNA1C, CACNA2D1, CACNB2, CALM1, CAV1, CBL, CBS, CCDC11, COL3A1, CRELD1, CRYAB, CSRP3, DMD, DNAH11, DNAH5, DSC2, DSG2, DSP, ENG, FBN2, FLNA, GATA6, GDF1, GDF2, GJA1, GJA5, GPD1L, HCN4, HRAS, JUP, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNK3, KCNQ1, KRAS, LEFTY2, LZTR1, MAP2K1, MAP2K2, MYH6, MYLK, MYPN, NAT10, NEBL, NEXN, NF1, NF2, NPPA, NRAS, NUP155, PKP2, PLN, PRKAG2, PTEN, PTPN11, RAF1, RANGRF, RASA1, RASA2, RBM20, RIT1, RRAS, RYR2, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SGCD, SHOC2, SHROOM3, SLC2A10, SLMAP, SMAD3, SNTA1, SOS1, SOS2, SPRED1, TBCK, TCAP, TGFB2, TGFB3, TMEM43, TNNC1, TNNI3, TPM1, TRDN, TRPM4, TSC1, TSC2, TTR, VCL*

*Only genes with a clinically proven relationship to cardiovascular diseases are accepted.

**Molecular Genetics Laboratory**

### Shipping Instructions

Please enclose a test requisition form with sample. All information must be complete before sample can be processed. Samples may be shipped at room temperature by overnight Federal Express to arrive Monday through Friday.

### Ship to:

**Molecular Genetics Lab**
Cincinnati Children's Hospital
3333 Burnet Ave. NRB 1042
Cincinnati, OH 45229

Phone: 513-636-4474
Fax: 513-636-4373

Updated 4/10/2017

## Methodology:

This test will perform DNA sequencing of the coding regions and their flanking 15 bp regions of the gene specified by the ordering physician. Sequencing over the promoter regions, deep intronic regions, or other regulatory elements will need to be specifically requested by the ordering physician.

## Analytical Sensitivity:

The sensitivity of DNA sequencing based test is over 99% for the detection of nucleotide base changes, small deletions, and small insertion in the region of interest. This assay will not detect variants in the promoter regions, deep intronic regions, or other regulatory elements unless specially requested by the ordering physician. This test does not detect large DNA rearrangements, deletions, insertions, or low level mosaicisms.

## References:


1. Grody W.W., Richards C.S. (2008). New quality assurance standards for rare disease testing. Genet Med 10 (5): 320-4.
2. Ledbetter D.H., Faucett W.A. (2008). Issues in genetic testing for ultra-rare diseases: background and introduction. Genet Med 10 (5): 309-13.
3. Maddalena A, Bale S., Das S., Grody W., Richards S. (2005). Technical standards and guidelines: molecular genetic testing for ultra-rare disorders. Genet Mrf 7(8):571-583.
4. Practice guidelines for Sanger sequencing analysis and interpretation. Prepared and edited by Ellard S., Carlton R., Yau M., Gokhale D., Taylor GR., Wallace A., and Ramsden SC. Ratified by the CMGS Executive Committee on 7th August 2009.


## Specimen:

Peripheral blood in EDTA tube
Adult: 3-5mL
Child: 3-5mL
Infant: 1-3mL
For other specimen types, please contact us at 513-636-4474

## Turnaround Time:

Single gene mutation analysis can take up to 90 days depending on the size of the gene.

## CPT Codes:

CPT coding is based on the gene being tested. Call for more information (513) 636-4474


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