THE MOLECULAR GENETICS DIAGNOSTIC LABORATORY AT CHILDREN’S HOSPITAL OF EASTERN ONTARIO IS PLEASED TO ANNOUNCE AN EXPANSION TO INHERITED CARDIOMYOPATHIES GENETIC TESTING SERVICE

Background information:
As of July 1, 2015, the Molecular Genetics Diagnostic Laboratory will be expanding the genetic testing panels for hypertrophic cardiomyopathy and arrhythmogenic right ventricular cardiomyopathy to include 19 genes and 7 genes, respectively. Additionally, we will be offering new testing panels for dilated cardiomyopathy (25 genes) and catecholaminergic polymorphic ventricular tachycardia (2 genes), as well as a pan-cardiomyopathy panel (45 genes). See page two for a list of the genes included in each panel.

Testing will be performed via sequencing and multiplex ligation-dependent probe amplification (MLPA). Sequencing consists of analysis of coding sequences of the relevant genes and 10 base pairs immediately adjacent to each exon. This test is performed by oligonucleotide-based target capture (TruSight Cardiomyopathy Panel, Illumina) followed by next generation sequencing using the MiSeq instrument (Illumina). Additional Sanger sequencing is performed for relevant regions that have insufficient coverage, and to confirm all clinically significant variants and variants of unknown significance.

Turn Around Time:
Full panel: 10 weeks
Variant-specific analysis: 3 to 4 weeks
Urgent analysis (must be discussed with the laboratory directly): 4 weeks

Sample Requirements: (room temperature)
Blood: 10 mL EDTA (lavender top), 3 mL EDTA for infants 1 year or less
For any other sample types, please contact the laboratory directly prior to sending the sample.

For more information and our requisition, visit http://cheo.on.ca/en/moleculargenetics or contact:

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INHERITED CARDIOMYOPATHIES TEST DETAILS

Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) Panel
Genes included in panel: DSC2, DSG2, DSP, JUP, PKP2, RYR2, and TMEM43
Analysis includes sequencing, and MLPA of DSC2, DSG2, DSP and PKP2.

Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) Panel
Genes included in panel: CASQ2 and RYR2
Analysis includes sequencing.

Hypertrophic Cardiomyopathy (HCM) Panel
Genes included in panel: ACTC1, ACTN2, CAV3, CSRP3, GLA, LAMP2, MYBPC3, MYH7, MYL2, MYL3, MYOZ2, NEXN, PLN, PRKAG2, TNNC1, TNNI3, TNNT2, TPM1, and TTR
Analysis includes sequencing, and MLPA of MYH7, MYBPC3 and TNNT2.

Dilated Cardiomyopathy (DCM) Panel
Genes included in panel: ABCC9, ACTC1, ACTN2, CSRP3, CTF1, DES, EMD, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, NEXN, PLN, RBM20, SGCD, TAZ, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTN, and VCL
Analysis includes sequencing, and MLPA of MYH7, MYBPC3 and TNNT2.

Pan Cardiomyopathy Panel
Genes included in panel: ABCC9, ACTC1, ACTN2, ANKRD1, CASQ2, CAV3, CRYAB, CSRP3, CTF1, DES, DSC2, DSG2, DSP, EMD, FHL2, GLA, JUP, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, NEXN, PKP2, PLN, PRKAG2, RBM20, RYR2, SGCD, TAZ, TCAP, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, and VCL
Analysis includes sequencing, and MLPA of DSC2, DSG2, DSP, MYH7, MYBPC3, PKP2 and TNNT2.