

Full name:	Cardiopathies, hereditary (102 genes) - KUL
Abbreviation:	HCMLQT
Description:	MLPA performed for the MYBPC3 gene (only in the context of hypertrophic cardiomyopathy).
Version number:	v3
Laboratory:	<u>Centrum Menselijke Erfelijkheid - KUL</u>
Created:	28 Jun 2019 - 09:13
Changed:	06 Sep 2022 - 16:05

Related Diseases

- ATTRV122I amyloidosis
- ATTRV30M amyloidosis
- Andersen-Tawil syndrome
- Atrial septal defect, ostium secundum type
- Atrial standstill
- Barth syndrome
- Brugada syndrome
- Carvajal syndrome
- Catecholaminergic polymorphic ventricular tachycardia
- Congenital fiber-type disproportion myopathy
- Congenital muscular dystrophy due to LMNA mutation
- Ebstein malformation of the tricuspid valve
- Erythrokeratoderma-cardiomyopathy syndrome
- Fabry disease
- Familial atrial fibrillation
- Familial bicuspid aortic valve
- Familial isolated arrhythmogenic ventricular dysplasia, biventricular form
- Familial isolated arrhythmogenic ventricular dysplasia, left dominant form
- Familial isolated arrhythmogenic ventricular dysplasia, right dominant form

- Familial isolated dilated cardiomyopathy
- Familial isolated restrictive cardiomyopathy
- Familial progressive cardiac conduction defect
- Familial short QT syndrome
- Familial sick sinus syndrome
- Familial thoracic aortic aneurysm and aortic dissection
- Fatal congenital hypertrophic cardiomyopathy due to glycogen storage disease
- Glycogen storage disease due to LAMP-2 deficiency
- Heart-hand syndrome, Slovenian type
- Hypoplastic left heart syndrome
- Idiopathic ventricular fibrillation, non Brugada type
- Jervell and Lange-Nielsen syndrome
- Late-onset distal myopathy, Markesbery-Griggs type
- Left ventricular noncompaction
- Muscular dystrophy, Selcen type
- Naxos disease
- Romano-Ward syndrome
- Sinoatrial node dysfunction and deafness
- Tetralogy of Fallot
- Timothy syndrome

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ABCC9</u>	95.00	0	NM_005691.4 / interpretable range CS1>95%
<u>ACTC1</u>	95.00	0	NM_005159.5 / interpretable range CS1>95%
<u>ACTN2</u>	95.00	0	NM_001103.4 / nterpretable range CS1>95%
<u>AKAP9</u>	95.00	0	NM_005751.5 /interpretable range CS1>95%
<u>ANK2</u>	95.00	0	NM_001148.6 / interpretable range CS1>95%
<u>ANKRD1</u>	95.00	0	NM_014391.3 / interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>BAG3</u>	95.00	0	NM_004281.4 / interpretable range CS1>95%
<u>CACNA1C</u>	95.00	0	NM_000719.7 / interpretable range CS1>95%
<u>CACNA1D</u>	95.00	0	NM_000720.4 / interpretable range CS1>95%
<u>CACNA2D1</u>	95.00	0	NM_000722.4 / interpretable range CS1>95%
<u>CACNB2</u>	95.00	0	NM_201590.3 / interpretable range CS1>95%
<u>CALM1</u>	95.00	0	NM_006888.6 / interpretable range CS1>95%
<u>CALM2</u>	95.00	0	NM_001743.6 / interpretable range CS1>95%
<u>CALM3</u>	95.00	0	NM_005184.4 / interpretable range CS1>95%
<u>CALR3</u>	95.00	0	NM_145046.5 / interpretable range CS1>95%
<u>CASQ2</u>	95.00	0	NM_001232.4 / interpretable range CS1>95%
<u>CAV3</u>	95.00	0	NM_033337.3 / interpretable range CS1>95%
<u>CDH2</u>	95.00	0	NM_001792.5 / interpretable range CS1>95%
<u>CFH</u>	95.00	0	NM_001449.5 / interpretable range CS1>95%
<u>CRYAB</u>	95.00	0	NM_001885.3 / interpretable range CS1>95%
<u>CSRP3</u>	95.00	0	NM_003476.5 / interpretable range CS1>95%
<u>CTNNA3</u>	95.00	0	NM_013266.4 / interpretable range CS1>95%
<u>DES</u>	95.00	0	NM_001927.4 / interpretable range CS1>95%
<u>DSC3</u>	95.00	0	NM_024422.6 / interpretable range CS1>95%
<u>DSG2</u>	95.00	0	NM_001943.5 / interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>DSP</u>	95.00	0	NM_004415.4 / interpretable range CS1>95%
<u>DTNA</u>	95.00	0	NM_001390.4 / interpretable range CS1>95%
<u>FHOD3</u>	95.00	0	NM_025135.5 / interpretable range CS1>95%
<u>FKTN</u>	95.00	0	NM_001079802.2 / interpretable range CS1>95%
<u>FLNC</u>	95.00	0	NM_001458.5 / interpretable range CS1>95%
<u>GJA5</u>	95.00	0	NM_005266.7 / interpretable range CS1>95%
<u>GLA</u>	95.00	0	NM_000169.3 / interpretable range CS1>95%
<u>GPD1L</u>	95.00	0	NM_015141.4 / interpretable range CS1>95%
<u>HCN4</u>	95.00	0	NM_005477.3 / interpretable range CS1>95%
<u>JPH2</u>	95.00	0	NM_020433.5 / interpretable range CS1>95%
<u>JUP</u>	95.00	0	NM_002230.4 / interpretable range CS1>95%
<u>KCNA5</u>	95.00	0	NM_002234.4 / interpretable range CS1>95%
<u>KCND2</u>	95.00	0	NM_012281.3 / interpretable range CS1>95%
<u>KCND3</u>	95.00	0	NM_004980.5 / interpretable range CS1>95%
<u>KCNE1</u>	95.00	0	NM_000219.6 / interpretable range CS1>95%
<u>KCNE2</u>	95.00	0	NM_172201.2 / interpretable range CS1>95%
<u>KCNE3</u>	95.00	0	NM_005472.5 / interpretable range CS1>95%
<u>KCNE5</u>	95.00	0	NM_012282.4 / interpretable range CS1>95%
<u>KCNH2</u>	95.00	0	NM_000238.4 / interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>KCNJ2</u>	95.00	0	NM_000891.3 / interpretable range CS1>95%
<u>KCNJ5</u>	95.00	0	NM_000890.5 / interpretable range CS1>95%
<u>KCNJ8</u>	95.00	0	NM_004982.4 / interpretable range CS1>95%
<u>KCNQ1</u>	95.00	0	NM_000218.3 / interpretable range CS1>95%
<u>LAMA4</u>	95.00	0	NM_002290.5 / interpretable range CS1>95%
<u>LAMP2</u>	95.00	0	NM_002294.3 / interpretable range CS1>95%
<u>LDB3</u>	95.00	0	NM_001080116.1 / interpretable range CS1>95%
<u>LMNA</u>	95.00	0	NM_170707.4 / interpretable range CS1>95%
<u>MIB1</u>	95.00	0	NM_020774.4 / interpretable range CS1>95%
<u>MYBPC3</u>	95.00	0	NM_000256.3 / interpretable range CS1>95% / MLPA only in de frame of "Familial hypertrophic cardiomyopathy"
<u>MYH6</u>	95.00	0	NM_002471.4 / interpretable range CS1>95%
<u>MYH7</u>	95.00	0	NM_000257.4 / interpretable range CS1>95%
<u>MYL2</u>	95.00	0	NM_000432.4 / interpretable range CS1>95%
<u>MYL3</u>	95.00	0	NM_000258.3 / interpretable range CS1>95%
<u>MYLK2</u>	95.00	0	NM_033118.4 / interpretable range CS1>95%
<u>MYOZ2</u>	95.00	0	NM_016599.5 / interpretable range CS1>95%
<u>MYPN</u>	95.00	0	NM_032578.4 / interpretable range CS1>95%
<u>NEXN</u>	95.00	0	NM_144573.4 / interpretable range CS1>95%
<u>NKX2-5</u>	95.00	0	NM_004387.4 / interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>NOS1AP</u>	95.00	0	NM_014697.3 / interpretable range CS1>95%
<u>NPPA</u>	95.00	0	NM_006172.4 / interpretable range CS1>95%
<u>NUP155</u>	95.00	0	NM_153485.3 / interpretable range CS1>95%
<u>PITX2</u>	95.00	0	NM_153427.2 / interpretable range CS1>95%
<u>PKP2</u>	95.00	0	NM_004572.4 / interpretable range CS1>95%
<u>PLN</u>	95.00	0	NM_002667.5 / interpretable range CS1>95%
<u>PRKAG2</u>	95.00	0	NM_016203.4 / interpretable range CS1>95%
<u>RBM20</u>	95.00	0	NM_001134363.3 / interpretable range CS1>95%
<u>RYR2</u>	95.00	0	NM_001035.3 / interpretable range CS1>95%
<u>SCN10A</u>	95.00	0	NM_006514.4 / interpretable range CS1>95%
<u>SCN1B</u>	95.00	0	NM_001037.5 / interpretable range CS1>95%
<u>SCN2B</u>	95.00	0	NM_004588.5 / interpretable range CS1>95%
<u>SCN3B</u>	95.00	0	NM_018400.4 / interpretable range CS1>95%
<u>SCN4B</u>	95.00	0	NM_174934.4 / interpretable range CS1>95%
<u>SCN5A</u>	95.00	0	NM_198056.3 / interpretable range CS1>95%
<u>SEMA3A</u>	95.00	0	NM_006080.3 / interpretable range CS1>95%
<u>SGCD</u>	95.00	0	NM_000337.6 / interpretable range CS1>95%
<u>SLC4A3</u>	95.00	0	NM_201574.2 / interpretable range CS1>95%
<u>SNTA1</u>	95.00	0	NM_003098.3 / interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>TBX20</u>	95.00	0	NM_001077653.2 / interpretable range CS1>95%
<u>TCAP</u>	95.00	0	NM_003673.4 / interpretable range CS1>95%
<u>TECRL</u>	95.00	0	NM_001010874.5 / interpretable range CS1>95%
<u>TGFB3</u>	95.00	0	NM_003239.5 / interpretable range CS1>95%
<u>TJP1</u>	95.00	0	NM_003257.5 / interpretable range CS1>95%
<u>TMEM43</u>	95.00	0	NM_024334.3 / interpretable range CS1>95%
<u>TMPO</u>	95.00	0	NM_003276.2 / interpretable range CS1>95%
<u>TNNI3</u>	95.00	0	NM_000363.5 / interpretable range CS1>95%
<u>TNNI3K</u>	95.00	0	NM_015978.3 / interpretable range CS1>95%
<u>TNNT2</u>	95.00	0	NM_001001430.3 / interpretable range CS1>95%
<u>TPM1</u>	95.00	0	NM_001018005.2 / interpretable range CS1>95%
<u>TRDN</u>	95.00	0	NM_006073.4 / interpretable range CS1>95%
<u>TRIM63</u>	95.00	0	NM_032588.4 / interpretable range CS1>95%
<u>TRPM4</u>	95.00	0	NM_017636.4 / interpretable range CS1>95%
<u>TTN</u>	95.00	0	NM_001267550.1 / interpretable range CS1>95%
<u>TTR</u>	95.00	0	NM_000371.4 / interpretable range CS1>95%
<u>TXNRD2</u>	95.00	0	NM_006440.5 / interpretable range CS1>95%
<u>VCL</u>	95.00	0	NM_014000.3 / interpretable range CS1>95%
<u>WWTR1</u>	95.00	0	NM_000116.5 / interpretable range CS1>95%