

Report panel PDF

Full name:	Cardiomyopathy, hereditary (208 genes) - VUB
Type of panel:	<u>Custom panel</u>
Provider:	home made
Laboratory:	<u>Centrum Medische Genetica - UZ Brussel VUB</u>
Created:	15 Jul 2019 - 14:00
Changed:	27 Sep 2022 - 15:46

Related Diseases

- ATTRV12I amyloidosis
- ATTRV30M amyloidosis
- Andersen-Tawil syndrome
- Atrial septal defect, ostium secundum type
- Atrial standstill
- Atypical hemolytic-uremic syndrome with H factor anomaly
- Autosomal dominant Emery-Dreifuss muscular dystrophy
- Barth syndrome
- Brugada syndrome
- Carvajal syndrome
- Catecholaminergic polymorphic ventricular tachycardia
- Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome
- Congenital fiber-type disproportion myopathy
- Costello syndrome
- Dilated cardiomyopathy with ataxia
- Early-onset myopathy with fatal cardiomyopathy
- Erythrokeratoderma-cardiomyopathy syndrome
- FKRP-related limb-girdle muscular dystrophy R9
- Fabry disease
- Familial atrial fibrillation
- Familial dilated cardiomyopathy with conduction defect due to LMNA mutation

- 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 congenital asplenia
- Familial isolated dilated cardiomyopathy
- Familial isolated restrictive cardiomyopathy
- Familial progressive cardiac conduction defect
- Familial short QT syndrome
- Familial thoracic aortic aneurysm and aortic dissection
- Fatal infantile cytochrome C oxidase deficiency
- Fatal infantile hypertonic myofibrillar myopathy
- Friedreich ataxia
- Glycogen storage disease due to LAMP-2 deficiency
- Glycogen storage disease due to acid maltase deficiency, infantile onset
- Late-onset distal myopathy, Markesberry-Griggs type
- Left ventricular noncompaction
- Leigh syndrome with leukodystrophy
- Naxos disease
- Noonan syndrome
- Noonan syndrome with multiple lentigines
- Romano-Ward syndrome
- Sensorineural deafness with dilated cardiomyopathy
- Very long chain acyl-CoA dehydrogenase deficiency
- X-linked Emery-Dreifuss muscular dystrophy

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
ABCC9	100.00	0	No comment
ACAD9	100.00	0	No comment
ACADVL	98.89	0	No comment
ACE	97.11	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ACTA1</u>	100.00	0	No comment
<u>ACTC1</u>	100.00	0	No comment
<u>ACTN2</u>	100.00	0	No comment
<u>ADD3</u>	100.00	0	No comment
<u>ADRB2</u>	100.00	0	No comment
<u>AGTR1</u>	100.00	0	No comment
<u>AGXT</u>	100.00	0	No comment
<u>AKAP9</u>	100.00	0	No comment
<u>ALPK3</u>	98.02	0	No comment
<u>ANKRD1</u>	100.00	0	No comment
<u>APOA1</u>	99.97	0	No comment
<u>B2M</u>	100.00	0	No comment
<u>BAG3</u>	100.00	0	No comment
<u>BRAF</u>	96.97	0	No comment
<u>CACNA1C</u>	100.00	0	No comment
<u>CACNB2</u>	100.00	0	No comment
<u>CALM1</u>	100.00	0	No comment
<u>CALM2</u>	100.00	0	No comment
<u>CALM3</u>	99.99	0	No comment
<u>CALR3</u>	100.00	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>CASQ2</u>	100.00	0	No comment
<u>CASZ1</u>	95.56	0	No comment
<u>CAV3</u>	100.00	0	No comment
<u>CAVIN4</u>	100.00	0	No comment
<u>CBL</u>	100.00	0	No comment
<u>CDH2</u>	97.48	0	No comment
<u>CFH</u>	100.00	0	No comment
<u>CHRM2</u>	100.00	0	No comment
<u>CMYA5</u>	99.99	0	No comment
<u>CNBP</u>	100.00	0	No comment
<u>COA5</u>	100.00	0	No comment
<u>COA6</u>	100.00	0	No comment
<u>COX10</u>	100.00	0	No comment
<u>COX15</u>	100.00	0	No comment
<u>CRYAB</u>	100.00	0	No comment
<u>CSRP3</u>	100.00	0	No comment
<u>CTF1</u>	30.96	0	No comment
<u>CTNNA3</u>	100.00	0	No comment
<u>DES</u>	100.00	0	No comment
<u>DLG1</u>	100.00	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>DMD</u>	100.00	0	No comment
<u>DNAJC19</u>	100.00	0	No comment
<u>DNM1L</u>	100.00	0	No comment
<u>DOLK</u>	100.00	0	No comment
<u>DSC3</u>	98.44	0	No comment
<u>DSG2</u>	99.61	0	No comment
<u>DSP</u>	100.00	0	No comment
<u>DTNA</u>	100.00	0	No comment
<u>ECRG4</u>	99.32	0	No comment
<u>ELAC2</u>	99.99	0	No comment
<u>ELN</u>	100.00	0	No comment
<u>EMD</u>	99.84	0	No comment
<u>EYA4</u>	100.00	0	No comment
<u>FBXO32</u>	100.00	0	No comment
<u>FHL2</u>	100.00	0	No comment
<u>FHOD3</u>	100.00	0	No comment
<u>FKRP</u>	98.68	0	No comment
<u>FKTN</u>	100.00	0	No comment
<u>FLNC</u>	100.00	0	No comment
<u>FLT1</u>	99.53	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>FOXD4</u>	100.00	0	No comment
<u>FXN</u>	89.89	0	No comment
<u>GAA</u>	100.00	0	No comment
<u>GATA4</u>	80.69	0	No comment
<u>GATA5</u>	97.28	0	No comment
<u>GATA6</u>	81.11	0	No comment
<u>GATAD1</u>	88.18	0	No comment
<u>GLA</u>	100.00	0	No comment
<u>GTPBP3</u>	100.00	0	No comment
<u>HACD1</u>	95.46	0	No comment
<u>HADHB</u>	100.00	0	No comment
<u>HAND1</u>	100.00	0	No comment
<u>HCN4</u>	92.35	0	No comment
<u>HIF1A</u>	100.00	0	No comment
<u>HRAS</u>	100.00	0	No comment
<u>HSPB6</u>	99.65	0	No comment
<u>HSPB7</u>	100.00	0	No comment
<u>ILK</u>	100.00	0	No comment
<u>INS-IGF2</u>	99.94	0	No comment
<u>ISL1</u>	100.00	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>JPH2</u>	95.10	0	No comment
<u>JUP</u>	100.00	0	No comment
<u>KARS1</u>	100.00	0	No comment
<u>KAT2B</u>	94.80	0	No comment
<u>KCND2</u>	100.00	0	No comment
<u>KCNE1</u>	100.00	0	No comment
<u>KCNH2</u>	94.44	0	No comment
<u>KCNJ12</u>	100.00	0	No comment
<u>KCNQ1</u>	91.07	0	No comment
<u>KIF20A</u>	100.00	0	No comment
<u>KLF10</u>	100.00	0	No comment
<u>LAMA2</u>	100.00	0	No comment
<u>LAMA4</u>	100.00	0	No comment
<u>LAMP2</u>	100.00	0	No comment
<u>LDB3</u>	100.00	0	No comment
<u>LMNA</u>	99.93	0	No comment
<u>LRRC10</u>	100.00	0	No comment
<u>MAP2K1</u>	100.00	0	No comment
<u>MAP2K2</u>	99.98	0	No comment
<u>MED12</u>	100.00	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>MIB1</u>	100.00	0	No comment
<u>MIB2</u>	99.31	0	No comment
<u>MRPL3</u>	100.00	0	No comment
<u>MRPL44</u>	100.00	0	No comment
<u>MTO1</u>	95.75	0	No comment
<u>MYBPC3</u>	100.00	0	No comment
<u>MYH15</u>	100.00	0	No comment
<u>MYH6</u>	100.00	0	No comment
<u>MYH7</u>	100.00	0	No comment
<u>MYH7B</u>	99.96	0	No comment
<u>MYL2</u>	100.00	0	No comment
<u>MYL3</u>	100.00	0	No comment
<u>MYLK2</u>	100.00	0	No comment
<u>MYO6</u>	100.00	0	No comment
<u>MYOM1</u>	100.00	0	No comment
<u>MYOM3</u>	100.00	0	No comment
<u>MYOZ1</u>	100.00	0	No comment
<u>MYOZ2</u>	100.00	0	No comment
<u>MYPN</u>	100.00	0	No comment
<u>NAA10</u>	96.03	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>NCOA6</u>	100.00	0	No comment
<u>NDUFAF1</u>	100.00	0	No comment
<u>NDUFV2</u>	100.00	0	No comment
<u>NEB</u>	99.99	0	No comment
<u>NEBL</u>	100.00	0	No comment
<u>NEXN</u>	100.00	0	No comment
<u>NKX2-5</u>	100.00	0	No comment
<u>NPPA</u>	100.00	0	No comment
<u>NRAP</u>	100.00	0	No comment
<u>OBSCN</u>	99.26	0	No comment
<u>OBSL1</u>	98.31	0	No comment
<u>PDLIM3</u>	100.00	0	No comment
<u>PKP2</u>	99.87	0	No comment
<u>PLEC</u>	99.82	0	No comment
<u>PLEKHM2</u>	99.60	0	No comment
<u>PLN</u>	100.00	0	No comment
<u>PPCS</u>	100.00	0	No comment
<u>PRDM16</u>	100.00	0	No comment
<u>PRKAG2</u>	95.88	0	No comment
<u>PRNP</u>	100.00	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
PSEN1	100.00	0	No comment
PSEN2	100.00	0	No comment
PTEN	98.86	0	No comment
PTPN11	98.80	0	No comment
RAF1	100.00	0	No comment
RANGRF	100.00	0	No comment
RBM20	99.82	0	No comment
RIT1	100.00	0	No comment
RPSA	97.87	0	No comment
RRAGC	99.99	0	No comment
RTKN2	100.00	0	No comment
RYR2	100.00	0	No comment
S100A1	100.00	0	No comment
SCN4B	100.00	0	No comment
SCN5A	100.00	0	No comment
SDHA	98.02	0	No comment
SGCB	94.58	0	No comment
SGCD	100.00	0	No comment
SGCG	100.00	0	No comment
SGK1	99.99	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>SHOC2</u>	100.00	0	No comment
<u>SLC12A1</u>	100.00	0	No comment
<u>SLC22A5</u>	100.00	0	No comment
<u>SLC25A3</u>	100.00	0	No comment
<u>SLC25A4</u>	99.09	0	No comment
<u>SMC1A</u>	100.00	0	No comment
<u>SNTA1</u>	84.25	0	No comment
<u>SOD2</u>	100.00	0	No comment
<u>SOS1</u>	100.00	0	No comment
<u>SYNE1</u>	100.00	0	No comment
<u>SYNE2</u>	100.00	0	No comment
<u>SYNM</u>	92.59	0	No comment
<u>TAF1A</u>	100.00	0	No comment
<u>TAX1BP3</u>	100.00	0	No comment
<u>TBX20</u>	100.00	0	No comment
<u>TBX5</u>	100.00	0	No comment
<u>TCAP</u>	100.00	0	No comment
<u>TCF21</u>	100.00	0	No comment
<u>TGFB3</u>	100.00	0	No comment
<u>TJP1</u>	99.14	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>TK2</u>	95.90	0	No comment
<u>TMED4</u>	100.00	0	No comment
<u>TMEM43</u>	100.00	0	No comment
<u>TMEM87B</u>	99.37	0	No comment
<u>TMPO</u>	99.87	0	No comment
<u>TNNI3</u>	100.00	0	No comment
<u>TNNI3K</u>	100.00	0	No comment
<u>TNNT2</u>	100.00	0	No comment
<u>TNNT3</u>	100.00	0	No comment
<u>TPM1</u>	99.98	0	No comment
<u>TRIM54</u>	99.66	0	No comment
<u>TRIM55</u>	100.00	0	No comment
<u>TRIM63</u>	100.00	0	No comment
<u>TRPM4</u>	100.00	0	No comment
<u>TSFM</u>	93.22	0	No comment
<u>TTN</u>	100.00	0	No comment
<u>TTR</u>	100.00	0	No comment
<u>TXNRD2</u>	96.90	0	No comment
<u>VCL</u>	100.00	0	No comment
<u>VEGFA</u>	98.73	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
WWTR1	99.81	0	No comment
YWHAE	98.69	0	No comment
ZBTB17	100.00	0	No comment