Cardiac Diseases 202 genes

Analysis of all known genes associated with genetic cardiac diseases. Enables an accurate diagnosis.

State of the art diagnostics

- Large panel approach: **202 genes** that are associated with hereditary Cardiac Diseases are sequenced by next-generation sequencing (NGS)
- Differential diagnosis: Based on phenotype, any number of gene sets can be selected for interpretation
- **16 gene sets** available (see list on back)
- Very frequent updates – currently version 9
- Flexible sample material: EDTA blood, DNA, or dried blood spot cards
- **Sensitivity > 99.4 %**; Specificity > 99.9 %
- Highly automated laboratory workflow
- Successfully applied to thousands of patients
- Includes all known disease causing intronic and mtDNA variants
- Interpretation includes single nucleotide variants (SNVs), small insertions and deletions (INDELs), and copy number variants (CNVs) of single and/or multiple exons

Quality Made in Germany

- Comprehensive and easy to understand medical report with interpretation of the findings and recommendations
- Issued by an interdisciplinary team of scientists and medical doctors specialized in human genetics
- Fast results within 4-6 weeks
- Entire service performed in-house
- Neither samples nor data leave CeGaT
- Excellent price-performance-quality ratio

Case report

Patient: 56 year old male patient (age at diagnosis: 55 years)

Symptoms: Dilated cardiomyopathy (DD: viral myocarditis); disease progressed rapidly; soon after diagnosis patient had terminal heart failure.

Gene set interpreted: Cardiomyopathy, dilated (43 Genes, HRT01)

Result: Heterozygous pathogenic frameshift variant in gene TTN, encoding Titin (the largest human protein). TTN can only be analyzed secure and fast by NGS. Additionally, a published variant of uncertain significance (VUS) in TTR was identified that could contribute to the DCM of the patient.

Consequences: In this patient, a genetic (not viral) cause of DCM was proven by panel analysis and the patient was listed for cardiac transplantation. Molecular genetic testing regarding the identified pathogenic variants was offered to first-degree relatives. The early detection of mutation carriers is important to identify individuals who would benefit from surveillance, preventive measures and treatment in order to improve survival and quality of life.

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Genes & gene sets

Cardiomyopathy, dilated (53 Genes, HRT01)
ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, CASQ2, CRYAB, CSRP3, DES, DMD, DSC2, DSG2, DSP, DTNA, EMD, FKTN, FLNC, GATAD1, ILK, JPH2, JUP, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYPN, NEBL, NEXN, PDLIM3, PKP2, PLN, PRDM16, RAF1, RBM20, RYR2, SCN5A, SDHA, SGCD, TAZ, TCAP, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, VCL

Cardiomyopathy, hypertrophic (35 Genes, HRT02)
ACTC1, ACTN2, AKAP9, ANKRD1, CALR3, CAV3, CRYAB, CSRP3, DES, FHL1, FLNC, GAA, GLA, JPH2, LAMP2, LDB3, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, MYPN, NEXN, PLN, PRKAG2, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, VCL

Left Ventricular Noncompaction Cardiomyopathy (NCCM/LVNC) (14 Genes, HRT03)
ACTC1, DTNA, HCN4, LDB3, LMNA, MIB1, MYBPC3, MYH7, PKP2, PRDM16, TAZ, TNNT2, TPM1, TTN

Atrial Fibrillation and Short QT Syndrome (22 Genes, HRT04)
ABCC9, CACNA1C, CACNA2D1, CACNB2, GJA5, HCN4, KCNA5, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, LMNA, MYL4, NPPA, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, TBX5, TNNI3

Long QT Syndrome (19 Genes, HRT05)
AKAP9, ANK2, CACNA1C, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, RYR2, SCN4B, SCN5A, SNTA1, TECRL, TRDN

Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy (ARVD/C) (14 Genes, HRT06)
CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LMNA, PKP2, PLN, RYR2, TGFB3, TMEM43, TTN

Brugada-Syndrome (18 Genes, HRT07)
ABCC9, CACNA1C, CACNA2D1, CACNB2, GPD1L, HCN4, KCNAB2, KCND3, KCNE3, KCNH2, KCNJ8, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN5A, TRPM4

Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT); Paroxysmal/Idiopathic Ventricular Fibrillation / Tachycardia (14 Genes, HRT08)
ANK2, CALM1, CALM2, CALM3, CASQ2, GNAI2, HCN4, KCNJ2, KCNQ1, LMNA, RYR2, SCN5A, TECRL, TRDN

Isolated and syndromal congenital heart defects (83 Genes, HRT09)
ACTA2, ACTC1, ACVR2B, AFF4, BMPR2, CAD, CCDC11, CDK13, CELSR1, CELSR2, CELSR3, CFC1, CHD4, CHD7, CITED2, CREBBP, CRELD1, DNAH11, DNAH5, DNAH6, DNAI1, DTNA, EHMT1, ELN, EVC, EVC2, FBN1, FLNA, FOXC1, FOXH1, GANAB, GATA4, GATA5, GATA6, GDF1, GJA1, GPC3, HAND1, HAND2, HRAS, JAG1, KDM5B, KMT2D, LEFTY2, MED13L, MMP21, MYH11, MYH6, MYH7, NIPBL, NKX2-5, NKX2-6, NME7, NODAL, NOTCH1, NOTCH2, NR2F2, PITX2, PKD1L1, PLD1, POGZ, PRDM6, PRKD1, RABGAP1L, RBFOX2, RBM10, SALL4, SEMA3D, SEMA3E, SMAD6, TAB2, TBX1, TBX20, TBX5, TFAP2B, TGFBR1, TGFBR2, TLL1, TMEM260, TPM1, ZEB2, ZFPM2, ZIC3

RASopathies (30 Genes, HRT10)
AKT3, BRAF, CBL, CCND2, EPHB4, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NF1, NF2, NRAS, PIK3CA, PIK3R2, PPP1CB, PTPN11, RAF1, RASA1, RASA2, RIT1, RRAS, SASH1, SHOC2, SMARCB1, SOS1, SOS2, SPRED1, STAMBP

Aortenaneurysma / Loeys-Dietz-Syndrome / Arterial Tortuosity Syndrom (HRT11)
Is replaced by **CTD02: Connective Tissue Diseases** (Ehlers-Danlos Syndrome, Marfan Syndrome, Loeys-Dietz Syndrome, Aortic Aneurysm and Differential Diagnoses); please use the „Order Form ‚Connective Tissue Disease'".

Cardiomyopathy, restrictive (14 Genes, HRT12)
ACTC1, BAG3, CRYAB, DES, FLNC, MYBPC3, MYH7, MYL2, MYL3, MYPN, TNNI3, TNNT2, TPM1, TTR

Cardiomyopathy with onset in neonatal period, infancy or childhood (53 Genes, HRT13)
AARS2, ABCC9, ACAD9, ACADVL, ACTA1, ACTC1, AGK, ALMS1, ALPK3, BAG3, COA5, COA6, COX15, CPT2, CRYAB, DNAJC19, DOLK, DSP, ELAC2, FLNC, GAA, GBE1, GLA, GTPBP3, HADHA, HADHB, HRAS, KARS, LAMP2, MRPL3, MRPL44, MTO1, MYBPC3, MYH7, MYPN, NDUFB11, NDUFV2, PPA2, PRKAG2, RAF1, SCO2, SDHA, SLC22A5, SLC25A20, SLC25A3, SLC25A4, TAZ, TK2, TMEM70, TNNI3, TNNT2, TPM1, TSFM

Neuromuscular diseases with cardiomyopathy (43 Genes, HRT14)
AACTA1, AGL, BAG3, CAV3, CHKB, CRYAB, DES, DMD, DYSF, EMD, ETFA, ETFB, ETFDH, FHL1, FKRP, FKTN, FLNC, GAA, GBE1, KARS, LAMA2, LAMP2, LDB3, LIMS2, LMNA, MYH7, MYOT, NEB, PNPLA2, POMT1, POMT2, SCO2, SGCA, SGCB, SGCD, SGCG, SLC22A5, SYNE1, TAZ, TCAP, TOR1AIP1, TTN, VCP

Pulmonary arterial hypertension (11 Genes, HRT15)
ACTC1, BAG3, CRYAB, DES, FLNC, MYBPC3, MYH7, MYL2, MYL3, MYPN, TNNI3, TNNT2, TPM1, TTR

Hypercholesterolemia and Hyperlipoproteinemia (11 Genes, HRT16)
ABCA1, ABCG5, ABCG8, APOA5, APOB, APOE, LDLR, LDLRAP1, LIPA, LPL, PCSK9

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