REQUISITION FORM FOR GENETIC DISEASES AND PHARMACOGENETICS
CARDIOLOGY

REFFERING DOCTOR (MANDATORY):

Name (or print label): _____ ID number: _____

Hospital: _____ Service: _____

E-mail: _____ Telephone: _____

Signature: Date: _____/_____/_____

Do you authorize the report to be sent by e-mail? Yes ☐ No ☐

If yes, please write your institutional e-mail address**:** _____

PATIENT IDENTIFICATION (MANDATORY):

Name: _____ Gender: F ☐ M ☐

Identification number: _____ Date of birth: _____/_____/_____

GenoMed use only:

Post label(s)

Conferred by:

CLINICAL DATA AND DIAGNOSIS*

Affected ☐ **Not affected (Asymptomatic)** ☐ Ideal expected date: _____/_____/_____

*Attach, whenever possible and can be justified, any relevant clinical information, family data including consanguinity, other cases in the family, family tree, etc.

FAMILIAL INFORMATION

Index case (affected) ☐ **Familial variant*** ☐ **Spouse** ☐

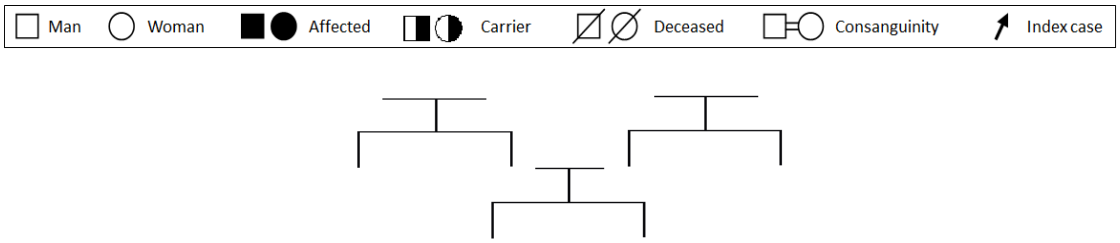
Known familial variant? NO ☐ YES ☐

If yes*: Gene/RefSeq: _____/_____ Variant: _____

Was the index case studied at GenoMed? NO ☐ YES ☐

***For carrier studies and predictive testing (familial variant study), please attach a copy of the index case, whenever possible and applicable.
According to the Portuguese law, (article 9º, law nº 12/2005), carrier studies and predictive testing (conducted in healthy individuals) it is ESSENTIAL that the clinical geneticist makes the request and that the patient informed consent is obtained.

Information for Pedigree construction:



SAMPLE

Blood (EDTA) ☐ DNA ☐ Tumor ☐ _____% (infiltration percentage)

Other (specify) ☐ _____

COLLECTION

Date: _____/_____/_____

Hour: _____:_____

INFORMED CONSENT (to be filled by the referring doctor)

I hereby declare that the patient informed consent for diagnosis was obtained. YES ☐ NO ☐

I hereby declare that the patient informed consent for investigation was obtained. YES ☐ NO ☐

CONTACT PERSONS: Dr.ª Diana Antunes, MD (dianaantunes@medicina.ulisboa.pt) / Dr. Yuri Chiodo, PhD (ychiodo@medicina.ulisboa.pt) Ext. 47301/48308

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REQUISITION FORM FOR GENETIC DISEASES AND PHARMACOGENETICS
CARDIOLOGY

GENETIC TEST REQUIRED*

☐ **Disease/Gene(s)**: _____

☐ **Study for familial variant** *(specify at "familial information")*

☐ **NGS genetic panel, with CNVs analysis** *(panel name)*: _____

☐ Additional genes *(if you want to personalize the above panel please specify which genes do you want to add or remove)*: _____

_____

_____

_____

☐ **Additional NGS genes analysis, with CNVs analysis** (based on total exome) for:

☐ NGS panel *(panel name)*: _____

☐ Gene(s) *(specify)*: _____

_____

_____

_____

☐ **DNA extraction**

☐ **Other studies***: _____

_____

*** Consult our website. If the test you want is not listed, please contact us.**

INFORMED CONSENT (mandatory – to be filled by the patient)

I hereby declare that I have read the Privacy Policy of GenoMed® - Diagnósticos de Medicina Molecular, S.A., available at https://genomed.pt/en/privacy-and-cookies-policy/ and I give my consent to the processing of the personal data. ☐ Agree ☐ Not agree

I hereby declare to authorize the collection of mine/my _____ [affiliation], _____ [name], born on _____/_____/_____, to the execution of the genetic testing described above, whose purposes and limitations were explained by the aforementioned physician. Herewith I declare that I have been informed about the consequences resulting from the teste results. I agree that the sample may be stored in order to allow repetition of the tests or further related tests at GenoMed or authorized partners around the world. The data/test results are subject to medical confidentiality and should only be disclosed to family members or other physicians with my permission. I am entitled to revoke this consent at any time.

☐ Agree ☐ Not agree

I also declare that the data/test results may be used in scientific investigations and publications in an anonymized form when and only approved by the Ethics Committee.

☐ Agree ☐ Not agree

(According to the Direction of General Health Standard nº 015/2013 updated.)

Patient's signature: _____ **Date:** _____/_____/_____

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Cardiomyopathies

34900	**Cardiomyopathies - panel of 169 genes by NGS:** *A2ML1, AARS2, ABCC9, ACADVL, ACTA1, ACTC1, ACTN2, AGL, AKAP9, ALG10, ALMS1, ALPK3, ANK2, ANKRD1, ATP5E, BAG3, BRAF, CACNA1C, CALM1, CALR3, CASQ2, CAV3, CAVIN4, CBL, CDH2, CHRM2, COA5, CPT2, CRYAB, CSRP3, CTF1, CTNNA3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSG3, DSP, DTNA, ELAC2, EMD, EPG5, EYA4, FHL1, FHL2, FHOD3, FKRP, FKTN, FLNC, FOXRED1, FXN, GAA, GATA4, GATA6, GATAD1, GLA, GLB1, GPD1L, GUSB, HAMP, HCN4, HFE, HFE2, HRAS, IDH2, ILK, JPH2, JUP, KCND3, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNQ1, KLF10, KRAS, LAMA4, LAMP2, LDB3, LMNA, LRRC10, MAP2K1, MAP2K2, MED12, MIB1, MRPL3, MTO1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYO6, MYOM1, MYOZ1, MYOZ2, MYPN, NEBL, NEXN, NF1, NKX2-5, NPPA, NRAS, OBSCN, PDLIM3, PKP2, PLEKHM2, PLN, PPA2, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, RAB3GAP2, RAF1, RASA1, RBM20, RIT1, RPL3L, RRAS, RYR2, SCN1B, SCN3B, SCN4B, SCN5A, SCO2, SDHA, SGCA, SGCB, SGCD, SHOC2, SLC22A5, SLC25A3, SLC25A4, SLC40A1, SNTA1, SOS1, SOS2, SPRED1, SYNE1, SYNE2, TAZ, TCAP, TFR2, TGFB3, TJP1, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRIM63, TRPM4, TSFM, TTN, TTR, TXNRD2, VCL, XK*	O
34900	**Arrhythmogenic cardiomyopathy - panel of 19 genes by NGS:** *CASQ2, CDH2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LMNA, MIB1, PKP2, PLN, RYR2, SCN5A, TGFB3, TJP1, TMEM43, TTN*	O
34900	**Left ventricular noncompaction - panel of 16 genes by NGS:** *ACTC1, ACTN2, DTNA, FHL2, FHOD3, ILK, LAMP2, LMNA, LDB3, MIB1, MYBPC3, MYH7, PRDM16, TAZ, TNNT2, TPM1*	O
34900	**Left ventricular noncompaction / Barth syndrome:** *TAZ* gene	O

Dilated Cardiomyopathy

34900	**Dilated cardiomyopathy - extended panel of 101 genes by NGS:** *ABCC9, ACADVL, ACTA1, ACTC1, ACTN2, ALMS1, ANKRD1, BAG3, CAV3, CAVIN4, CHRM2, CPT2, CRYAB, CSRP3, CTF1, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, EMD, EPG5, EYA4, FHL1, FHL2, FHOD3, FKRP, FKTN, FLNC, GATA4, GATA6, GATAD1, GLA, HAMP, HCN4, HFE, HFE2, IDH2, ILK, JPH2, JUP, KCND3, LAMA4, LAMP2, LDB3, LMNA, LRRC10, MED12, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYOZ1, MYPN, NEBL, NEXN, NKX2-5, NPPA, PDLIM3, PKP2, PLEKHM2, PLN, PRDM16, PRKAG2, PSEN1, PSEN2, RAB3GAP2, RAF1, RBM20, RPL3L, RYR2, SCN1B, SCN5A, SDHA, SGCA, SGCB, SGCD, SLC22A5, SLC40A1, SOS1, SYNE1, SYNE2, TAZ, TCAP, TFR2, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, TTR, TXNRD2, VCL, XK*	O
34900	**Dilated cardiomyopathy - basic panel of 35 genes by NGS:** *ACTC1, ACTN2, ANKRD1, BAG3, CSRP3, DES, DSG2, DMD, DSP, EMD, EYA4, FLNC, JPH2, LDB3, LMNA, MYBPC3, MYH6, MYH7, NEXN, PLN, PSEN1, PSEN2, RBM20, RPL3L, SCN5A, SGCD, TAZ, TCAP, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TTN, VCL*	O
34900	**Dilated cardiomyopathy:** *ACTC1* gene	O
34666	**Dilated cardiomyopathy:** *LMNA* gene	O
36352	**Dilated cardiomyopathy:** *MYH7* gene	O
36353	**Dilated cardiomyopathy:** *TNNT2* gene	O
34900	**Dilated cardiomyopathy:** *TPM1* gene	O
34900	**Dilated cardiomyopathy:** *MYH7, TNNT2, TPM1, ACTC1* genes	O

Hypertrophic Cardiomyopathy

34900	**Hypertrophic cardiomyopathy (sarcomeric genes) - panel of 8 genes by NGS:** *ACTC1, MYBPC3, MYH7, MYL2, MYL3, TNNI3, TNNT2, TPM1*	O
34900	**Hypertrophic cardiomyopathy - extended panel of 86 genes by NGS:** *A2ML1, AARS2, ACADVL, ACTA1, ACTC1, ACTN2, AGL, ALPK3, ANKRD1, ATP5E, BAG3, BRAF, CACNA1C, CALR3, CASQ2, CAV3, CBL, COA5, CPT2, CRYAB, CSRP3, DES, ELAC2, FHL1, FHOD3, FLNC, FOXRED1, FXN, GAA, GATA4, GLA, GLB1, GUSB, HRAS, JPH2, KCNQ1, KLF10, KRAS, LAMP2, LDB3, LMNA, MAP2K1, MAP2K2, MRPL3, MTO1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYO6, MYOM1, MYOZ2, MYPN, NEXN, NF1, NRAS, OBSCN, PDLIM3, PLN, PRKAG2, PTPN11, RAF1, RASA1, RIT1, RRAS, RYR2, SCO2, SHOC2, SLC25A3, SLC25A4, SOS1, SOS2, SPRED1, TCAP, TMEM70, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TSFM, TTN, TTR, VCL*	O
34900	**Hypertrophic cardiomyopathy - basic panel of 26 genes by NGS:** *ACTC1, ACTN2, ALPK3, CSRP3, GLA, KRAS, LAMP2, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYOZ2, NEXN, PLN, PRKAG2, PTPN11, RAF1, RIT1, SOS1, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTR*	O
34900	**Hypertrophic cardiomyopathy:** *ACTC1* gene	O
34900	**Hypertrophic cardiomyopathy:** *CSRP3* gene	O
34900	**Hypertrophic cardiomyopathy:** *FHL1* gene	O
36351	**Hypertrophic cardiomyopathy:** *MYBPC3* gene	O
36352	**Hypertrophic cardiomyopathy:** *MYH7* gene	O
34900	**Hypertrophic cardiomyopathy:** *MYL2* gene	O
34900	**Hypertrophic cardiomyopathy:** *MYL3* gene	O
36354	**Hypertrophic cardiomyopathy:** *TNNI3* gene	O
36353	**Hypertrophic cardiomyopathy:** *TNNT2* gene	O
34900	**Hypertrophic cardiomyopathy:** *TPM1* gene	O
34727	**Fabry disease:** *GLA* gene - index case	O
34726	**Fabry disease:** *GLA* gene - familial case	O

Arrhythmia / Cardiac Conduction Diseases

34900	**Cardiac arrhythmias - panel of 202 genes by NGS:** *A2ML1, AARS2, ABCC9, ACAD9, ACADVL, ACTA1, ACTC1, ACTN2, AGK, AGL, AGPAT2, AKAP9, ALMS1, ANK2, ANKRD1, ANO5, ATP5E, ATPAF2, BAG3, BRAF, BSCL2, CACNA1C, CACNA1D, CACNA2D1, CALM1, CALM2, CALM3, CALR3, CAPN3, CASQ2, CAV3, CAVIN4, CDH2, CHRM2, COA5, COA6, COQ2, COX15, COX6B1, CRYAB, CSRP3, CTNNA3, DES, DLD, DMD, DNAJC19, DOLK, DPP6, DSC2, DSG2, DSG3, DSP, DTNA, ELAC2, EMD, EYA4, FAH, FHL1, FHL2, FHOD3, FKRP, FKTN, FLNC, FOXRED1, FXN, GAA, GATA4, GATA6, GATAD1, GFM1, GJA1, GJA5, GLA, GLB1, GNPTAB, GPD1L, GUSB, HCN4, HRAS, JPH2, JUP, KCNA1, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNK17, KCNK3, KCNQ1, KCNQ2, KCNT1, KRAS, LAMA2, LAMA4, LAMP2, LDB3, LDLR, LIAS, LMNA, LZTR1, MAP2K1, MAP2K2, MIB1, MLYCD, MRPL3, MRPL44, MRPS22, MTO1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYL4, MYOM1, MYOT, MYOZ2, MYPN, NEBL, NEXN, NF1, NKX2-5, NKX2-6, NNT, NPPA, NRAS, OBSCN, PDHA1, PDLIM3, PHKA1, PITX2, PKP2, PLN, PMM2, PPA2, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, RAF1, RANGRF, RASA2, RBM20, RIT1, RRAS, RYR2, SCN1A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCN9A, SCO2, SDHA, SEMA3A, SGCD, SHOC2, SLC22A5, SLC25A3, SLC25A4, SLMAP, SNTA1, SOS1, SOS2, SPEG, SPRED1, SURF1, SYNE1, SYNE2, TAZ, TBX20, TBX5, TCAP, TGFB3, TJP1, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TOR1AIP1, TPM1, TRDN, TRIM63, TRPM4, TSFM, TTN, TTR, TXNRD2, VCL, XK*	O
34900	**Sudden death - panel of 299 genes by NGS:** *A2ML1, AARS2, ABCA1, ABCA12, ABCC6, ABCC9, ABCG5, ABCG8, ACAD9, ACADVL, ACTA1, ACTA2, ACTC1, ACTN2, AGK, AGL, AGPAT2, AKAP9, AKT1, ALG10, ALG10B, ALMS1, ALPK3, ANK2, ANKRD1, ANO5, APOA2, APOA5, APOB, APOC2, APOC3, APOE, APTX, ATP5E, ATPAF2, BAG3, BRAF, BSCL2, CACNA1C, CACNA1D, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CALR3, CAPN3, CASQ2, CAV3, CAVIN4, CBL, CBS, CDH2, CHRM2, CLCF1, COA5, COA6, COL3A1, COL5A1, COL5A2, COQ2, COX15, COX6B1, CPT1A, CPT2, CREB3L3, CRLF1, CRYAB, CSRP3, CTF1, CTNNA3, CYP27A1, DEPDC5, DES, DLD, DMD, DNAJC19, DOLK, DPP6, DSC2, DSG2, DSG3, DSP, DTNA, EFEMP2, ELAC2, ELN, EMD, ENPP1, EPG5, EPHX2, EYA4, FAH, FBN1, FBN2, FGF12, FHL1, FHL2, FHOD3, FKRP, FKTN, FLNA, FLNC, FOXRED1, FXN, GAA, GATA4, GATA5, GATA6, GATAD1, GFM1, GHR, GJA1, GJA5, GLA, GLB1, GNAI2, GNPTAB, GPD1, GPD1L, GPIHBP1, GUSB, HAMP, HCN4, HFE, HFE2, HRAS, IDH2, IKZF1, ILK, ITIH4, JPH2, JUP, KCNA1, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNK17, KCNK3, KCNQ1, KCNQ2, KCNQ3, KCNT1, KLF10, KRAS, LAMA2, LAMA4, LAMP2, LDB3, LDLR, LDLRAP1, LIAS, LIPA, LIPI, LMF1, LMNA, LPL, LRP6, LRRC10, LZTR1, MAP2K1, MAP2K2, MAT2A, MED12, MFAP5, MIB1, MLYCD, MRPL3, MRPL44, MRPS22, MTO1, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYL4, MYLK, MYLK2, MYO6, MYOM1, MYOT, MYOZ1, MYOZ2, MYPN, NEBL, NEXN, NF1, NKX2-5, NKX2-6, NNT, NOTCH1, NPPA, NRAS, OBSCN, PCDH19, PCSK9, PDHA1, PDLIM3, PHKA1, PITX2, PKP2, PLEKHM2, PLN, PLOD1, PMM2, PPA2, PPP1R17, PRDM16, PRKAG2, PRKG1, PRRT2, PSEN1, PSEN2, PTPN11, RAB3GAP2, RAF1, RANGRF, RASA1, RASA2, RBM20, RIT1, RRAS, RPL3L, RYR2, SCN10A, SCN1A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCN8A, SCN9A, SCO2, SDHA, SEMA3A, SGCA, SGCB, SGCD, SHOC2, SKI, SLC22A5, SLC25A3, SLC25A4, SLC2A1, SLC2A10, SLC40A1, SLMAP, SMAD3, SMAD4, SMAD6, SNTA1, SOS1, SOS2, SPEG, SPRED1, SURF1, SYNE1, SYNE2, TAZ, TBX20, TBX5, TCAP, TFR2, TGFB2, TGFB3, TGFBR1, TGFBR2, TJP1, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TNXB, TOR1AIP1, TPM1, TRDN, TRIM63, TRPM4, TSFM, TTN, TTR, TXNRD2, USF1, VCL, XK*	O
34900	**Brugada syndrome - panel of 42 genes by NGS:** *ABCC9, AKAP9, ANK2, ANK3, CACNA1C, CACNA1D, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CAV3, DPP6, FGF12, GPD1L, HCN4, IRX5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, PKP2, PXDNL, RANGRF, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SEMA3A, SLMAP, SNTA1, TRDN, TRPM4*	O

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34900	**Brugada syndrome:** *SCN5A* gene	O
34900	**Long QT syndrome - panel of 17 genes by NGS:** *AKAP9, ANK2, CACNA1C, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1, TRDN*	O
34900	**Long QT syndrome:** *KCNQ1, KCNH2, SCN5A* genes	O
34900	**Long QT syndrome (LQT1):** *KCNQ1* gene	O
34900	**Long QT syndrome (LQT2):** *KCNH2* gene	O
34900	**Long QT syndrome (LQT3):** *SCN5A* gene	O
34900	**Long QT syndrome (LQT5):** *KCNE1* gene	O
34900	**Catecholaminergic polymorphic ventricular tachycardia - panel of 8 genes by NGS:** *ANK2, CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TRDN*	O

Aortopathies / Conjunctivopathies

34900	**Aortic/Connective tissue diseases - basic panel of 64 genes by NGS:** *ABL1, ACTA2, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL4, ATP7A, B3GALT6, B3GLCT, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL11A1, COL11A2, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL9A1, COL9A2, DSE, EFEMP2, ELN, FBN1, FBN2, FKBP14, FLCN, FLNA, FOXE3, GAA, HRAS, IPO8, KCNJ8, LOX, LTBP2, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRDM5, PRKG1, PTPN11, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, TGFB2, TGFB3, TGFBR1, TGFBR2, TNXB, ZNF469*	O
34900	**Aortic/Connective tissue diseases - extended panel of 121 genes by NGS:** *ABCC6, ABL1, ACTA2, ACVR1, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL4, AEBP1, ALDH18A1, ALPL, ARHGAP31, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B3GAT3, B3GLCT, B4GALT7, BGN, BMP1, BMP4, C1R, C1S, CBS, CHST14, COL11A1, COL11A2, COL12A1, COL18A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, COL9A1, COL9A2, COL9A3, COX7B, CREB3L1, CRTAP, DCC, DLL4, DOCK6, DSE, EFEMP2, ELN, EMILIN1, EOGT, FBLN5, FBN1, FBN2, FKBP10, FKBP14, FLCN, FLNA, FOXE3, GAA, GGCX, GZF1, IPO8, GORAB, GYPC, HRAS, IFITM5, KCNJ8, KIF22, LOX, LRP5, LTBP2, LTBP4, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, P3H1, PIEZO2, PLOD1, PLOD2, PLOD3, PLS3, PPIB, PRDM5, PRKG1, PTPN11, PYCR1, RBPJ, RET, RIN2, ROBO3, SERPINF1, SERPINH1, SGMS2, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, SP7, SPARC, TAB2, TGFB2, TGFB3, TGFBR1, TGFBR2, TMEM38B, TNXB, TPSAB1, VCAN, WNT1, ZNF469*	O
34900	**Aortic diseases - panel of 31 genes by NGS:** *ACTA2, CBS, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBN1, FBN2, FLNA, GATA5, IPO8, LOX, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, SKI, SLC2A10, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, TNXB*	O
34900	**Ehlers-Danlos syndrome - panel of 22 genes by NGS:** *ADAMTS2, AEBP1, ATP7A, B3GALT6, B4GALT7, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, DSE, FKBP14, FLNA, KIF22, PLOD1, PRDM5, SCN9A, SLC39A13, TNXB, ZNF469*	O
34900	**Marfan and Marfan-like syndrome - panel of 9 genes by NGS:** *COL3A1, FBN1, FBN2, SKI, SLC2A10, SMAD3, TGFB2, TGFBR1, TGFBR2*	O
34900	**Aneurysm/Thoracic aortic dissection:** *ACTA2* gene	O
34900	**Ehlers-Danlos syndrome, classic form:** *COL5A1* gene	O
34900	**Ehlers-Danlos syndrome, vascular type:** *COL3A1* gene	O
34900	**Loeys-Dietz syndrome:** *TGFBR1* gene	O
34900	**Loeys-Dietz syndrome:** *TGFBR2* gene	O
34900	**Marfan syndrome:** *FBN1* gene	O

Other pathologies

34900	**Cardiomyopathy and arrhythmia - panel of 228 genes by NGS:** *A2ML1, AARS2, ABCC9, ACAD9, ACADVL, ACTA1, ACTC1, ACTN2, AGK, AGL, AGPAT2, AKAP9, ALG10, ALMS1, ALPK3, ANK2, ANKRD1, ANO5, ATP5E, ATPAF2, BAG3, BRAF, BSCL2, CACNA1C, CACNA1D, CACNA2D1, CALM1, CALM2, CALM3, CALR3, CAPN3, CASQ2, CAV3, CAVIN4, CBL, CDH2, CHRM2, COA5, COA6, COQ2, COX15, COX6B1, CPT2, CRYAB, CSRP3, CTF1, CTNNA3, DES, DLD, DMD, DNAJC19, DOLK, DPP6, DSC2, DSG2, DSG3, DSP, DTNA, ELAC2, EMD, EPG5, EYA4, FAH, FHL1, FHL2, FHOD3, FKRP, FKTN, FLNC, FOXRED1, FXN, GAA, GATA4, GATA6, GATAD1, GFM1, GJA1, GJA5, GLA, GLB1, GNPTAB, GPD1L, GUSB, HAMP, HCN4, HFE, HFE2, HRAS, IDH2, ILK, JPH2, JUP, KCNA1, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNK17, KCNK3, KCNQ1, KCNQ2, KCNT1, KLF10, KRAS, LAMA2, LAMA4, LAMP2, LDB3, LDLR, LIAS, LMNA, LRRC10, LZTR1, MAP2K1, MAP2K2, MED12, MIB1, MLYCD, MRPL3, MRPL44, MRPS22, MTO1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYL4, MYLK2, MYO6, MYOM1, MYOT, MYOZ1, MYOZ2, MYPN, NEBL, NEXN, NF1, NKX2-5, NKX2-6, NNT, NPPA, NRAS, OBSCN, PDHA1, PDLIM3, PHKA1, PITX2, PKP2, PLEKHM2, PLN, PMM2, PPA2, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, RAB3GAP2, RAF1, RANGRF, RASA1, RASA2, RBM20, RIT1, RPL3L, RRAS, RYR2, SCN1A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCN9A, SCO2, SDHA, SEMA3A, SGCA, SGCB, SGCD, SHOC2, SLC22A5, SLC25A3, SLC25A4, SLC40A1, SLMAP, SNTA1, SOS1, SOS2, SPEG, SPRED1, SURF1, SYNE1, SYNE2, TAB2, TAZ, TBX20, TBX5, TCAP, TFR2, TGFB3, TJP1, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TOR1AIP1, TPM1, TRDN, TRIM63, TRPM4, TSFM, TTN, TTR, TXNRD2, VCL, XK*	O
34900	**Noonan syndrome / Rasopathies - panel of 25 genes by NGS:** *A2ML1, BRAF, CBL, FGD1, HRAS, KAT6B, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA1, RASA2, RIT1, RRAS, RRAS2, SHOC2, SOS1, SOS2, SPRED1*	O
34900	**Ciliopathies - panel of 176 genes by NGS:** *ACVR2B, ADGRV1, AHI1, AIPL1, ALMS1, ANKS6, ARL13B, ARL6, ARMC4, ATXN10, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C21orf59, C2orf71, C5orf42, CC2D2A, CCDC103, CCDC114, CCDC151, CCDC172, CCDC28B, CCDC39, CCDC40, CCDC65, CCDC96, CCNO, CDH23, CENPF, CEP104, CEP120, CEP164, CEP290, CEP295, CEP41, CEP83, CFAP300, CFAP53, CFTR, CLRN1, CRB1, CRELD1, CRX, CSPP1, DCDC2, DEUP1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, DYNC2H1, EVC, EVC2, EXOC6B, EXOC8, FAM166B, FOXH1, GAS2L2, GAS8, GDF1, GLIS2, GUCY2D, HYDIN, HYLS1, IFT122, IFT140, IFT172, IFT27, IFT43, IFT80, IMPDH1, INPP5E, INVS, IQCB1, KCNJ13, KIAA0556, KIAA0586, KIF14, KIF7, LCA5, LEFTY2, LRAT, LRRC34, LRRC6, LZTFL1, MAPKBP1, MCIDAS, MKKS, MKS1, MRE11, MYO7A, NEK1, NEK8, NKX2-5, NME8, NODAL, NPHP1, NPHP3, NPHP4, OFD1, PCDH15, PDE6D, PIEZO2, PIH1D3, PKD2, PKHD1, RD3, RDH12, RPE65, RPGR, RPGRIP1, RPGRIP1L, RSPH1, RSPH3, RSPH4A, RSPH9, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SPAG1, SPATA7, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM17, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TRIM32, TSC1, TSC2, TTC21B, TTC23, TTC25, TTC6, TTC8, TULP1, UMOD, USH1C, USH1G, USH2A, VHL, WDPCP, WDR19, WDR34, WDR35, WDR60, WHRN, XPNPEP3, ZIC3, ZMYND10, ZNF423*	O
34900	**Heritable pulmonary arterial hypertension - panel of 11 genes by NGS:** *ACVRL1, BMPR1B, BMPR2, CAV1, CBLN2, EIF2AK4, ENG, FOXF1, KCNA5, KCNK3, SMAD9*	O
34900	**Monogenic hypertension - panel of 27 genes by NGS:** *AOC1, ASIC3, CLCNKB, CUL3, CYP11B1, CYP11B2, CYP17A1, HSD11B2, KCNH2, KCNJ1, KLHL3, NOS3, NR3C2, PDE3A, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC12A1, SLC12A3, SCNN1B, SCNN1G, VHL, WNK1, WNK4*	O
34900	**Vascular and lymphatic malformations - panel of 63 genes by NGS:** *ACVRL1, ADAMTS13, ADAMTS3, AKT1, AKT3, ANTXR1, ATM, BMPR2, BRAF, CAV1, CBL, CCBE1, CCM2, DOCK6, ELMO2, ENG, EPHB4, FAT4, FLT4, FOXC2, GATA2, GDF2, GJC2, GLMN, GNAQ, KCNK3, KDR, KIF11, KRAS, KRIT1, LZTR1, MAP2K1, MAP3K3, MTOR, NF1, NF2, NRAS, PDCD10, PDGFRB, PIEZO1, PIK3CA, PTEN, PTPN11, RAF1, RASA1, RIT1, SHOC2, SMAD2, SMAD3, SMAD4, SMARCB1, SOS1, SOX18, SPRED1, STAMBP, TEK, TGFB2, TGFB3, TGFBR1, TGFBR2, TSC1, TSC2, VEGFC*	O
34900	**Angiotensin converting enzyme (*ACE*) I/D polymorphism genotype**	O

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