REQUISITION FORM FOR GENETIC DISEASES AND PHARMACOGENETICS
CARDIOLOGY

GENOMED
Está nos genes.

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:** _____

PATIENTE IDENTIFICATION (mandatory or past label)

Name:	Gender: F ☐ M ☐
Identification number:	Date of birth: _____/_____/_____

GenoMed use only:

Post label(s)

Conferred by:

CLINICAL DATA AND DIAGNOSIS*

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.

FAMILY INFORMATION

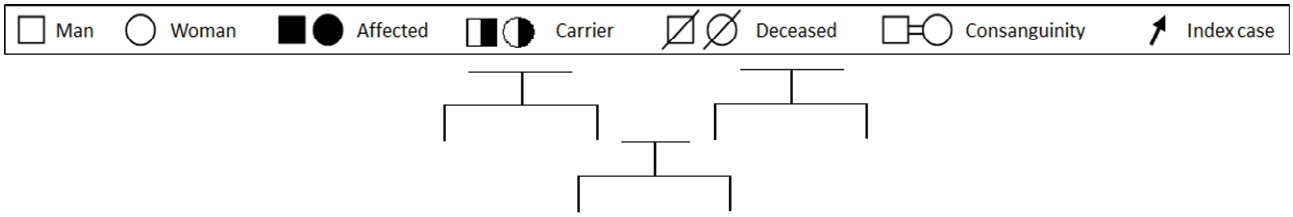
Index case (affected) ☐	**Familial case (affected)** ☐	**Familial case (asymptomatic)** ☐
Known familial variant?	NO ☐	YES ☐

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	COLLECTION
Blood (EDTA) ☐ DNA ☐ Tumor ☐ _____% (infiltration percentage)	Date: ____/_____/_____
Other (specify) _____	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 ☐

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

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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GENETIC TEST REQUIRED

☐ **Disease/Gene(s)**:

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

☐ **NGS genetic panel** *(panel name)*:

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

☐ With CNVs analysis*
* Additional cost.

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

☐ NGS panel *(panel name)*:

☐ Gene(s) *(specify)*:

☐ **DNA extraction**

☐ **Other studies**

Cardiomyopathies

34900	**Cardiomyopathies - panel of 103 genes by NGS:** *AARS2, ABCC9, ACTA1, ACTC1, ACTN2, ANKRD1, BAG3, CALR3, CASQ2, CAV3, CAVIN4, CHRM2, COA5, CRYAB, CSRP3, CTF1, CTNNA3, DES, DMD, DOLK, DSC2, DSG2, DSP, DTNA, EMD, EYA4, FHL1, FHL2, FHOD3, FKTN, FLNC, FOXRED1, FXN, GAA, GATAD1, GLA, GLB1, GUSB, HRAS, ILK, JPH2, JUP, KCNQ1, KLF10, KRAS, LAMA4, LAMP2, LDB3, LMNA, MAP2K1, MAP2K2, MIB1, MRPL3, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ1, MYOZ2, MYPN, NEBL, NEXN, NRAS, OBSCN, PDLIM3, PKP2, PLN, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, RAF1, RBM20, RIT1, RYR2, SCN5A, SCO2, SGCA, SGCD, SHOC2, SLC25A3, SLC25A4, SOS1, TAZ, TCAP, TGFB3, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRDN, TRIM63, TSFM, TTN, TTR, VCL*	O
34900	**Arrhythmogenic cardiomyopathy - panel of 17 genes by NGS:** *CASQ2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LMNA, MIB1, PKP2, PLN, RYR2, SCN5A, TGFB3, 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 - panel of 47 genes by NGS:** *ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, CRYAB, CSRP3, CTF1, DES, DMD, DSG2, DSP, EMD, EYA4, FHL2, FHOD3, FKTN, FLNC, GATAD1, ILK, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYOZ1, MYPN, NEXN, PKP2, PLN, PSEN1, PSEN2, RBM20, SCN5A, SGCA, SGCD, TAZ, TCAP, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TTN, VCL*	O
34900	**Dilated cardiomyopathy - panel of 32 genes by NGS:** *ACTC1, ACTN2, ANKRD1, BAG3, CSRP3, DES, DSG2, DMD, DSP, EMD, EYA4, LDB3, LMNA, MYBPC3, MYH6, MYH7, NEXN, PLN, PSEN1, PSEN2, RBM20, 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 - panel of 68 genes by NGS:** *AARS2, ACTA1, ACTC1, ACTN2, ANKRD1, BAG3, CALR3, CASQ2, CAV3, COA5, CRYAB, CSRP3, DES, FHL1, FHOD3, FLNC, FOXRED1, FXN, GAA, GLA, GLB1, GUSB, HRAS, JPH2, KCNQ1, KLF10, KRAS, LAMP2, LDB3, LMNA, MAP2K1, MAP2K2, MRPL3, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, NRAS, OBSCN, PDLIM3, PLN, PRKAG2, PTPN11, RAF1, RIT1, RYR2, SCO2, SHOC2, SLC25A3, SLC25A4, SOS1, TCAP, TMEM70, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TSFM, TTN, TTR, VCL*	O
34900	**Hypertrophic cardiomyopathy - panel of 25 genes by NGS:** *CTC1, ACTN2, 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 189 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, CACNB2, CALM1, CALM2, CALM3, CALR3, CAPN3, CASQ2, CAV3, CAVIN4, CHRM2, COA5, COA6, COQ2, COX15, COX6B1, CRYAB, CSRP3, CTNNA3, DES, DLD, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, DTNA, ELAC2, EMD, EYA4, FAH, FHL1, FHL2, FHOD3, FKRP, FKTN, FLNC, FOXD4, FOXRED1, FXN, GAA, GATA4, GATA6, GATAD1, GFM1, GJA1, GJA5, GLA, GLB1, GNPTAB, GPD1L, GUSB, HCN4, HRAS, JPH2, JUP, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNK17, KCNQ1, KRAS, LAMA2, LAMA4, LAMP2, LDB3, LDLR, LIAS, LMNA, LZTR1, MAP2K1, MAP2K2, MIB1, MLYCD, MRPL3, MRPL44, MRPS22, MTO1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYOM1, MYOT, MYOZ2, MYPN, NEBL, NEXN, NF1, NKX2-5, NKX2-6, NNT, NRAS, OBSCN, PDHA1, PHKA1, PITX2, PKP2, PLN, PMM2, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, RAF1, RANGRF, RASA2, RBM20, RIT1, RRAS, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCO2, SDHA, SGCD, SHOC2, SLC22A5, SLC25A3, SLC25A4, SLMAP, SNTA1, SOS1, SOS2, SPEG, SPRED1, SURF1, SYNE1, SYNE2, TAZ, TBX20, TBX5, TCAP, TGFB3, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TOR1AIP1, TPM1, TRDN, TRIM63, TRPM4, TSFM, TTN, TTR, TXNRD2, VCL, XK*	O
34900	**Sudden death without structural cardiopathy - panel of 65 genes by NGS:** *ABCC9, ACTC1, AKAP9, ANK2, CACNA1C, CACNA1D, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CAVIN4, DES, DSC2, DSG2, DSP, EMD, FGF12, FHL2, FLNC, GAA, GJA5, GLA, GPD1L, HCN4, JUP, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNK17, KCNQ1, LAMP2, LMNA, NKX2- 5, PKP2, PLN, PRKAG2, RANGRF, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SLMAP, SNTA1, TBX5, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TRDN, TRPM4, TTR*	O
34900	**Brugada syndrome - panel of 31 genes by NGS:** *ANK2, ANK3, ABCC9, CACNA1C, CACNA1D, CACNA2D1, CACNB2, CAV3, CLASP2, DPP6, FGF12, GPD1L, HCN4, IRX5, KCND2, KCND3, KCNE3, KCNE5, KCNH2, KCNJ8, PKP2, PXDNL, RANGRF, SCN1B, SCN2B, SCN3B, SCN5A, SCN10A, SEMA3A, SLMAP, TRPM4*	O

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34900	**Brugada syndrome:** *SCN5A* gene	O
34900	**Long QT syndrome - panel of 15 genes by NGS:** *AKAP9, ANK2, CACNA1C, CALM1, CALM2, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1*	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 - panel of 62 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, KCNJ8, LOX, LTBP2, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRDM5, PRKG1, PTPN11, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, TGFB2, TGFB3, TGFBR1, TGFBR2, ZNF469*	O
34900	**Aortic diseases - panel of 28 genes by NGS:** *ACTA2, CBS, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBN1, FBN2, FLNA, GATA5, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, SKI, SLC2A10, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2*	O
34900	**Ehlers-Danlos syndrome - panel of 20 genes by NGS:** *ADAMTS2, ATP7A, B3GALT6, B4GALT7, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, DSE, FKBP14, FLNA, KIF22, PLOD1, SLC39A13, TNXB, ZNF469, PRDM5, SCN9A*	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 196 genes by NGS:** *A2ML1, AARS2, ABCC9, ACAD9, ACADVL, ACTA1, ACTN2, AGK, AGL, AGPAT2, AKAP9, ALMS1, ANK2, ANKRD1, ANO5, ATP5E, ATPAF2, BAG3, BRAF, BSCL2, CACNA1C, CACNA1D, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CALR3, CAPN3, CASQ2, CAV3, CAVIN4, CHRM2, COA5, COA6, COQ2, COX15, COX6B1, CRYAB, CSRP3, CTF1, CTNNA3, DES, DLD, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, DTNA, ELAC2, EMD, EYA4, FAH, FGF12, FHL1, FHL2, FHOD3, FKRP, FKTN, FLNC, FOXD4, FOXRED1, FXN, GAA, GATA4, GATA6, GATAD1, GFM1, GJA1, GJA5, GLA, GLB1, GNPTAB, GPD1L, GUSB, HCN4, HRAS, ILK, JPH2, JUP, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNK17, KCNQ1, KLF10, KRAS, LAMA2, LAMA4, LAMP2, LDB3, LDLR, LIAS, LMNA, LZTR1, MAP2K1, MAP2K2, MIB1, MLYCD, MRPL3, MRPL44, MRPS22, MTO1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOT, MYOZ1, MYOZ2, MYPN, NEBL, NEXN, NF1, NKX2-5, NKX2-6, NNT, NRAS, OBSCN, PDHA1, PDLIM3, PHKA1, PITX2, PKP2, PLN, PMM2, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, RAF1, RANGRF, RASA2, RBM20, RIT1, RRAS, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCO2, SDHA, SGCA, SGCD, SHOC2, SLC22A5, SLC25A3, SLC25A4, SLMAP, SNTA1, SOS1, SOS2, SPEG, SPRED1, SURF1, SYNE1, SYNE2, TAZ, TBX20, TBX5, TCAP, TGFB3, 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 23 genes by NGS:** *A2ML1, BRAF, CBL, FGD1, HRAS, KAT6B, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1*	O
34900	**Ciliopathies - panel of 174 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, 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, 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	**Angiotensin converting enzyme (*ACE*) I/D polymorphism genotype**	O

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