

SEND TO

Genome Diagnostics Section
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UMC Utrecht

Laboratory Opening Hours :8:30-17:00 Mon-Fri

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Email genoomdiagnostiek@umcutrecht.nl

PATIENT DETAILS (complete in capitals or place patient sticker in box) Use one form per patient

Surname + initials/forename :

Address :

Postcode/residence :

Country :

Date of birth (DoB, DD/MM/YY) :

Sex :

BILLING DETAILS (complete in capitals)

.....

.....

REFERRING PHYSICIAN (complete in capitals)

Name (in full) : Date (DD/MM/YY) :

Hospital (in full) : Telephone :

Address : Email address :

Postcode/residence : Your reference (if applicable) :

Country : Copy report to (if applicable) :

TEST REQUIRED

- Indicate the desired gene panel analysis and/or individual gene analysis (see table from page 4 onwards) or include details of known familial mutation below.
- Include pedigree, clinical information and, if relevant, details of familial mutation and name and DoB of proband, on page 2 of this form.

Urgent, only after consultation. Please contact us by phone or email. Use courier delivery address to send sample(s) (see page 3).

PURPOSE

- Confirmation of clinical diagnosis
- Carrier testing (include details of familial mutation)
- Presymptomatic testing (include details of familial mutation)
- Partner testing
- Prenatal testing (**only after consultation**)
- DNA storage only (for possible future testing)
- Research (**only after consultation**)

FAMILY HISTORY

- Mutation unknown → indicate required test(s) in table from page 3 onwards
 - Familial mutation known → indicate relevant clinical information and proband relation to index patient in pedigree on page 2
- Gene :
- Mutation :
- Family number :
- Reference :

SAMPLE INFORMATION

Ensure patient sample tubes/vials are clearly labelled with **name, gender, DoB and time/date of collection**. We reserve the right to refuse to process samples with incomplete or ambiguous patient information. Has your patient received an **allogeneic hematopoietic stem cell transplant**? See page 3 for additional instructions. For sampling instructions and despatch/transfer procedures, see page 3.

- Blood* (2 x 10 mL EDTA, minimum 2 x 2 mL for neonates)
 - Chorionic villi (15 mg) (**only after consultation**)
 - Amniotic fluid (30 mL) (**only after consultation**)
 - Umbilical cord blood (5 mL)
 - Blood for RNA isolation (2 x 2,5 mL PAXgene blood tubes) (**only after consultation**)
 - Bone marrow | Tube type: EDTA Sodium Heparin
 - Tissue (2x 10 µg) | Type : Sample ID(s) :
 - Re-analysis existing data
- DNA (2x >10 µg) | Sample ID(s) :
- DNA sample in storage at the UMCU Genome Diagnostics laboratory

For all samples
 Date (DD/MM/YY) / time of collection:

USE OF BODY MATERIAL

By sending their body material for diagnostic testing, a person is effectively included as a patient of the UMC Utrecht. The UMC Utrecht uses residual human tissue to develop new and improve existing techniques and for further research in line with the original diagnostic request. The referring physician should inform the patient about this. (see page 3 and the patient information sheet for more information)

* see page 3

GENOME DIAGNOSTICS LABORATORY USE ONLY

U-nummer

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Datum:

Etiketten

Registratie

Indicatie:

Gericht / Volledig

Paraaf:

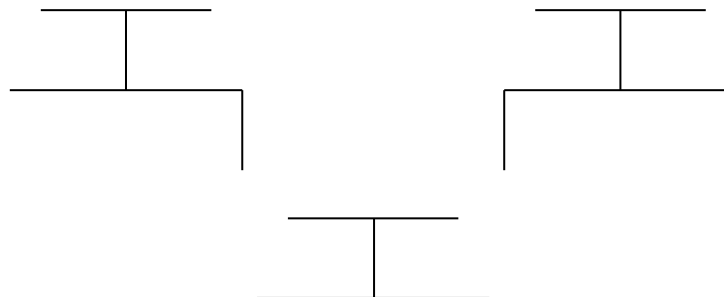
Ontvangstdatum

CLINICAL INDICATIONS:

Include relevant clinical information, pedigree, details of familial mutation and name and date of birth (DoB, DD/MM/YY) of proband if relevant.

PEDIGREE

Indicate patient with an arrow (→); use ■/● for affected, include name and DoB for all relatives previously tested.



Number in pedigree	Name	Date of birth (DD/MM/YY)

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Our gene panels and single gene tests are subject to change, please ensure the **most recent version of this form** is used (see top right for version number and date). The most recent version of our referral form is available on: <http://www.umcutrecht.nl/aanvraagGenoom>. The composition of our current and previous (versions of) gene panels is available on: www.umcutrecht.nl/NGS.

Sampling procedures

- Store patient samples overnight at 4°C if required, do **NOT** freeze or expose to heat.
- Samples can be sent at room temperature. Sample and referral forms should be sent together.
- If a test is requested on **chorionic villi, amniotic fluid or umbilical cord blood** a maternal sample is required to allow maternal cell contamination testing. Please use a separate referral form for the maternal sample.
 - For sampling procedures, please consult: <http://www.umcutrecht.nl/aanvraagGenoom>.
- **Courier address:** UMC Utrecht, DBG afdeling Genetica, Lundlaan6, KC.04.084.2, 3584 EA Utrecht. **Deliver to:** receptie afdeling Genetica KC.04.084.2.
- * After an **allogeneic hematopoietic stem cell transplant** blood is no longer suitable for DNA analysis. Please contact our laboratory via +31 (0)88 – 75 54090 for more information and alternative options.

Gene tests not listed in this form

Custom gene (panel) testing based on NGS sequencing is available upon request, also for genes not included in the listed tests. Contact us for more information.

Use of body material

Body material will be stored and may be used for further (diagnostic) research on the patients' behalf, or - after anonymization - for the improvement of current and implementation of new methods/techniques (see the patient information sheet for more information). For detailed information about privacy and the protection of personal data, we refer to the website of the UMC Utrecht: Practical > Rights and regulations > Use of residual material. Permission to use residual material can also be changed here. (see <https://www.umcutrecht.nl/nl/Ziekenhuis/In-het-ziekenhuis/Regels-en-rechten/Gebruik-lichaamsmateriaal-medische-gegevens/Bezwaarformulier>)

Confidentiality

The confidentiality of data is guaranteed and secured by the UMC Utrecht guidelines. See www.umcutrecht.nl.



The genome diagnostics section has been certified with NEN-EN-ISO 15189:2012 by the Accreditation Council. The scope of accreditation number M001 can be seen on www.rva.nl.

Blood disorders, vascular disease and bone marrow failure

Gene panels (incl. WES based CNV analysis per gene)

Bone marrow failure (BMF01v22.1; 122 genes)

The following non-coding RNA genes are not included in this analysis: *RMRP* and *TERC*. Please contact us for diagnostic opportunities if there's a strong suspicion of involvement for these non-coding genes.

ABCB7, ABCD4, ABCG5, ABCG8, ACBD5, ACD, ACKR1, AK2, AMN, ANKRD26, AP3B1, ATR, BRCA2, BRIP1, CD40LG, CECR1, CLCN7, CLPB, CSF3R, CTC1, CTLA4, CUBN, CXCR4, CYCS, DDX41, DHFR, DKC1, DNAJC21, EFL1, EIF2AK3, ELANE, ERCC4, ERCC6L2, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FYB1, G6PC3, GATA1, GATA2, GBA, GFI1, GP1BA, GP1BB, GP9, GRHL2, HAX1, HOXA11, IVD, JAGN1, KLF1, LIG4, LYST, MASTL, MECOM, MP1G6B, MPL, MTR, MTRR, MYH9, MYSM1, NBEAL2, NHEJ1, NHP2, NOP10, OSTM1, PALB2, PARN, PLEKHM1, PRF1, RAB27A, RAC2, RBM8A, RPL11, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS29, RPS7, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SH2D1A, SLC19A2, SLC25A38, SLC46A1, SLX4, SRC, SRP72, STIM1, STK4, STN1, TAZ, TBXAS1, TCIRG1, TCN2, TERT, THPO, TINF2, TNFRSF11A, TNFSF11, TUBB1, UBE2T, USB1, VPS13B, VPS45, WAS, WRAP53

Diamond-Blackfan anemia (DBA01v22.1; 35 genes)

EPO, GATA1, HEATR3, RPL10, RPL10A, RPL11, RPL15, RPL17, RPL18, RPL19, RPL26, RPL27, RPL3, RPL31, RPL34, RPL35, RPL35A, RPL5, RPL8, RPL9, RPLP0, RPS10, RPS11, RPS14, RPS15A, RPS17, RPS19, RPS20, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, TSR2

Hereditary hemolytic anemia (EMS00v17.1; 46 genes)

ABCB6, ABCG5, ABCG8, ADA, AK1, ALAS2, ALDOA, ANK1, ATP11C, C15orf41, CD59, CDAN1, COL4A1, CYB5R3, EPB41, EPB42, G6PD, GATA1, GCLC, GPI, GPX1, GSR, GSS, HBA1, HBA2, HBB, HK1, KCNN4, KIF23, KLF1, NT5C3A, PFKM, PGD, PGK1, PGLS, PIEZO1, PKLR, RHAG, SEC23B, SLC2A1, SLC4A1, SPTA1, SPTB, TALDO1, TPI1, XK

Primary hemostasis (TRO02v22.1; 100 genes)

ABCG5, ABCG8, ACTN1, ACVRL1, ADRA2A, ADRA2B, ANKRD26, ANO6, AP3B1, APOLD1, BLOC1S3, BLOC1S5, BLOC1S6, CD36, CDC42, COL1A1, COL3A1, COL5A1, COL5A2, CYCS, DIAPH1, DTNBP1, ENG, EPHB2, ETV6, F2R, F2RL3, FBN1, FERMT3, FGA, FGB, FGG, FLI1, FLNA, FYB1, GALE, GATA1, GATA2, GBA, GFI1B, GNA12, GNA13, GNAI1, GNAI2, GNAQ, GNAS, GNAZ, GNE, GP1BA, GP1BB, GP6, GP9, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, IKZF5, ITGA2, ITGA2B, ITGB1, ITGB3, LYST, MASTL, MECOM, MLPH, MPL, MYH9, MYO5A, NBEAL2, P2RX1, P2RY1, P2RY12, PLA2G4A, PLAU, PLCB2, PLCB3, PLCG2, PRKACG, PTGS1, PTPRJ, RAB27A, RASGRP2, RBM8A, RGS2, RUNX1, SLFN14, SMPD1, SRC, STIM1, TBX2R, TBXAS1, THPO, TPM4, TUBB1, VIPAS39, VPS33B, VWF, WAS

Congenital secondary erythrocytosis (EMS01v21.1; 16 genes)

BPGM, CYB5R3, EGLN1, EGLN2, EPAS1, EPO, EPOR, HBA1, HBA2, HBB, HIF3A, OS9, PIEZO1, PKLR, SH2B3, VHL

Rendu Osler Weber syndrome (ROW01v22.1; 4 genes)

Including deletion/duplication test *ENG* & *ACVRL1*
ENG, ACVRL1, GDF2, SMAD4

Blood disorders and vascular disease

Single gene | Sequence analysis

- G6PD deficientie G6PD
- Haemophilia A, (HEMA)[§] F8[§]
- Von Willebrand Factor VWF

Cardiovascular disease

Gene panels (incl. WES based CNV analysis per gene)

Cardiomyopathy (CAR01v21.1; 42 genes)

Relevant clinical information

- Hypertrophic (HCM)
- Dilated (DCM)[®] + Conduction abn.
- Arrhythmogenic right ventricle (ARVD/C)
- Left ventricle non compaction (LVNC)
- Restrictive (RCM)

ACTC1, ACTN2, ALPK3, BAG3, CRYAB, CSRP3, DES, DMD, DSC2, DSG2, DSP, FHL1, FLNC, GLA, HCN4, JPH2, JUP, LAMP2, LMNA, MIB1, MYBPC3, MYH7, MYL2, MYL3, NEXN, PKP2, PLN, PRDM16, PRKAG2, RBM20, RYR2, SCN5A, TAZ, TCAP, TMEM43, TNNC1, TNNI3, TNNI2, TPM1, TTN, TTR, VCL

Cardiac conduction abnormalities (CAR03v23.1; 49 genes)

Relevant clinical information

- Sudden cardiac arrest
- Sudden unexplained death
- Arrhythmogenic right ventricle (ARVD/C)
- Brugada syndrome (BrS)
- Sick Sinus syndrome (SSS)
- Atrial standstill
- Catecholaminergic polymorphic VT's (CPVT)
- Short QT syndrome (SQT)
- Long QT syndrome (LQT)

ABCC9, AKAP9, ANK2, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, DES, DPP6, DSC2, DSG2, DSP, GJA5, GPD1L, HCN4, JUP, KCNA5, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, LAMP2, LMNA, MYL4, NKX2-5, NPPA, PKP2, PLN, PRKAG2, RYR2, SCN2B, SCN3B, SCN4B, SCN5A, SNTA1, TBX5, TECRL, TMEM43, TRDN, TRPM4, TTN

Copy number analysis: PKP2 KCNQ1/KCNH2

Congenital heart defects (CAR05v19.1; 55 genes)

Relevant clinical information

- Non-syndromal
 - ASD/VSD/DORV
 - Heterotaxy
 - Tetralogy of Fallot (TOF)
- Syndromal
 - Heterotaxy
 - Velocardiofacial/DiGeorge (DGS)
 - Oculo-Facio-Cardio Dental
 - Holt-Oram (HOS)
 - Alstrom (ALMS)
 - Alagille (AGS)
 - Wolff-Parkinson-White (WPW)
 - Cantú syndrome
 - Noonan/LEOPARD (NS/LS)
 - Cardio-Facio-Cutaneous (CFC)

ALMS1, ACTC1, ACVR2B, BRAF, CBL, CFAP53, CFC1, CHD7, CITED2, CRELD1, ELN, FOXH1, GATA4, GATA5, GATA6, GDF1, GJA1, GJC1, HAND1, HAND2, HRAS, JAG1, KRAS, LDB3, LEFTY2, MAP2K1, MAP2K2, MED13L, MMP21, MYBPC3, MYH11, MYH6, MYH7, NKX2-5, NKX2-6, NODAL, NOTCH1, NOTCH2, NR2F2, NRAS, PKD1L1, PTPN11, RAF1, SHOC2, SMAD6, SOS1, TAB2, TAZ, TBX1, TBX20, TBX5, TFAP2B, TLL1, ZFPM2, ZIC3

Copy number analysis: MYBPC3

Pulmonary Arterial Hypertension (PAH) (CAR08v22.1; 15 genes)

ABCC8, ACVRL1, ATP13A3, BMPR2, CAV1, EIF2AK4, ENG, GDF2, GGX4, KCNK3, KDR, SMAD9, SOX17, TBX4, TET2

In submitting this sample the clinician confirms that the patient has been informed about the chances of uncovering incidental findings that can result from this medical test.

[§] Sequence and copy number analysis

[^] Repeat expansion analysis only

Cardiovascular disease (continued)

Gene panels (incl. WES based CNV analysis per gene)

Vascular disorders (CAR04v23.1; 38 genes)

Relevant clinical information

- Familial thoracic aortic aneurysm and aortic dissection (TAAD)
- Marfan (MFS)
- Loeys-Dietz (LDS)

ACTA2, ARIH1, BGN, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBN1, FBN2, FLNA, FOXE3, GATA4, GATA5, HCN4, LMOD1, LOX, LTBP3, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, PRKG1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFB3, TGFB3, TGFB3, THSD4

Idiopathic VF / Sudden Cardiac Death (SCD) (CAR09v23.1; 43 genes)

ACTC1, ACTN2, BAG3, CACNA1C, CALM1, CALM2, CALM3, CASQ2, DES, DMD, DSC2, DSG2, DSP, DPP6, FLNC, JUP, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, LAMP2, LMNA, MYBPC3, MYH7, MYL2, MYL3, PKP2, PPA2, PLN, PRKAG2, RBM20, RYR2, SCN5A, SLC4A3, TECRL, TMEM43, TNNC1, TNNT1, TNNT2, TPM1, TRDN, TTN

Cardiovascular disease

Single gene | Sequence analysis

- Alveolar capillary dysplasia with misalignment of the pulmonary veins (ACDMPV) FOXF1
- Brugada syndrome SCN5A
- Fabry disease GLA
- Long QT syndrome, type 1 and 2 (copy number analysis only) KCNQ1/KCNH2
- Syndromaal microphthalmia 2 (MCOPS2) / Oculofaciocardiodentaal syndroom (OFCD) BCOR

Dysmorphology

Gene panels (incl. WES based CNV analysis per gene)

- Fraser syndrome (FRA00v16.1; 4 genes)**
FRAS1, FREM2, FREM1, GRIP1
- Hypodontia/Oligodontia (DON01v19.1; 17 genes)**
AXIN2, BCOR, EDA, EDAR, EDARADD, FGFR1, FLNA, GJA1, GREM2, IRF6, LRP6, LTBP3, MSX1, PAX9, TP63, WNT10A, WNT10B
- Amelogenesis imperfecta (DON02v19.1; 27 genes)**
ACPT, AMBN, AMELX, C4orf26, CNNM4, COL17A1, DLX3, ENAM, FAM20A, FAM20C, FAM83H, GPR68, ITGB6, KLK4, LAMA3, LAMB3, LTBP3, MMP20, ORAI1, PEX1, PEX6, RELT, ROGDI, SLC13A5, SLC24A4, STIM1, WDR72
- Hemifacial microsomia (OWS01v19.1; 43 genes)**
Includes copy number analysis of EYA1
BMP4, CDC6, CDT1, CHD7, DHODH, EDNRA, EFTUD2, EIF4A3, EYA1, FGF10, FGF3, FGFR2, FGFR3, FRAS1, FREM2, GNAI3, GRIP1, GSC, HMX1, HOXA2, HSPA9, KDM6A, KMT2D, OFD1, ORC1, ORC4, ORC6, OTX2, PLCB4, POLR1A, POLR1C, POLR1D, SALL1, SALL4, SF3B4, SIX1, SLC26A4, SOX10, TCOF1, TFAP2A, GDF6, RPS28, SIX5
- (Non)syndromaal cleft lip and/or palate incl. Robin sequence (OWS02v20.2; 195 genes)**
Pre-test genetic counselling required
ACTB, ACTG1, ALX1, ALX3, AMER1, AMMECR1, ANKRD11, ARHGAP29, ARHGAP31, ASXL1, B3GALT6, B3GALT7, B4GALT7, B9D2, BCOR, BMP2, BMPER, C2CD3, C5orf42, CC2D2A, CDC45, CDH1, CDKN1C, CHD7, CHRNG, CHST14, COL11A1, COL11A2, COL2A1, COL9A1, COLEC10, COLEC11, CTCF, CTNND1, DDX3X, DDX59, DHCR7, DHODH, DLL4, DOCK6, DVL1, DVL3, DYNC2H1, DYNC2L1, EBP, EDN1, EDNRA, EFNB1, EFTUD2, EIF2S3, EIF4A3, EOGT, EPG5, ESCO2, EYA1, FAM20C, FGD1, FGF8, FGFR1, FGFR2, FLNA, FLNB, FOXC2, FOXE1, FRAS1, FTO, GDF6, GJA1, GLI2, GLI3, GNAI3, GNB1, GPC3, GRHL3, HDAC8, HYL51, ICK, IFT140, IFT172, IFT57, IFT80, IMPAD1, INTU, IRF6, KANSL1, KAT6A, KCNJ2, KCNK9, KDM6A, KIAA0196, KIAA0586, KIAA1279, KIF7, KMT2D, MAP3K7, MAPRE2, MASP1, MBTPS2, MED25, MEIS2, MID1, MKS1, MSX1, NEDD4L,

NEK1, NIPBL, NOTCH1, OFD1, ORC1, PAX3, PGM1, PHF8, PHGDH, PIEZO2, PIGN, PIGV, PLCB4, POLR1A, POLR1C, POLR1D, POMT1, PORCN, PQBP1, PROKR2, PRRX1, PTCH1, PTCH2, PVRL1, RBM10, RIPK4, ROR2, RPRGRIPL, RPL11, RPL26, RPL5, RPS19, RPS26, RPS28, RUNX2, SALL4, SATB2, SCARF2, SEC23A, SEMA3E, SEPT9, SF3B4, SHH, SIX1, SIX3, SIX5, SKI, SLC26A2, SMAD3, SMAD4, SMC1A, SMC3, SMCHD1, SMS, SNRBP, SON, SOX9, SPECC1L, STAC3, STAMBP, TAPT1, TBX1, TBX15, TBX2, TBX22, TCOF1, TCTN3, TFAP2A, TGDS, TGFB3, TGFB3, TGFB3, TGFB3, TGFB3, TGMCO1, TMEM216, TMEM8C, TP63, TRIM37, TUBB, TWIST1, TXNL4A, USP9X, WDR35, WNT4, WNT5A, XYLT1, ZEB2, ZIC2, ZIC3, ZMPSTE24, ZSWIM6

Pierre Robin Sequence (OWS03v19.1; 20 genes)

AMER1, COL11A1, COL11A2, COL2A1, DHODH, EDN1, EFTUD2, GNAI3, PGM1, PLCB4, POLR1A, POLR1C, POLR1D, RBM10, SATB2, SF3B4, SLC26A2, SOX9, TBX1, TCOF1

Dysmorphology

Single gene | Sequence analysis

- Cantú syndrome ABCC9
- Cleidocranial dysplasia (CCD)[§] RUNX2[§]
- Currarino, triade van (TRIAD) MNX1
- Syndromaal microphthalmia 2 (MCOPS2) / Oculofaciocardiodentaal syndroom (OFCD) BCOR

Epilepsy

Gene panels (incl. WES based CNV analysis per gene)

Epilepsy full gene panel (EPI00v21.1; 301 genes)

AARS1, ABAT, ACTL6B, ADPRS, ADSL, ALDH7A1, ALG13, AMT, ANKRD11, AP2M1, AP3B2, ARG1, ARHGGEF9, ARV1, ARX, ASAH1, ATAD1, ATP1A1, ATP1A2, ATP1A3, ATP6AP2, ATP6V1A, ATRX, BRAT1, C12orf57, CACNA1A, CACNA1B, CACNA1D, CACNA1E, CACNA1G, CACNA2D2, CAD, CASK, CDK19, CDKL5, CERS1, CHD2, CHRNA2, CHRNA4, CHRN2, CIC, CLCN4, CLN3, CLN5, CLN6, CLN8, CLTC, CNKSR2, CNM2, CNPY3, CNTNAP2, COQ2, COQ4, CPLX1, CPT2, CSNK2B, CSTB, CTSD, CUL4B, CUX2, CYFIP2, D2HGDH, DCX, DEAF1, DENND5A, DEPDC5, DHDDS, DHPS, DIAPH1, DNM2, DNAJC5, DNMI1, DNMI2, DOCK7, DPM1, DYRK1A, EEF1A2, EIF2S3, EIF3F, EPM2A, FARS2, FGD1, FGF12, FLNA, FOLR1, FOXG1, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA3, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GAMT, GATM, GCSH, GLB1, GLDC, GLRA1, GLRB, GNAO1, GNB1, GNB5, GOSR2, GOT2, GPAA1, GPC3, GPHN, GRIA3, GRIA4, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRM7, GRN, HACE1, HCF1, HCN1, HECW2, HNRNP, HNRNP, HSD17B10, HUWE1, INTS8, IQSEC2, IRF2BPL, KANSL1, KARS1, KAT8, KCNA1, KCNA2, KCNB1, KCNC1, KCNC2, KCND3, KCNH1, KCNJ10, KCNM1A, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD3, KCTD7, KDM5C, KEXMIF, KIF1A, KMT2A, KPNA7, LGI1, LIAS, MAST3, MBD5, MBOAT7, MDH2, MECP2, MED12, MEF2C, MFSDB, MOCS1, MOCS2, MPDU1, MTHFR, MTOR, NACCC1, NABP, NBEA, NHLRC1, NPLRL2, NPLRL3, NFXN1, NSDHL, NTRK2, NUS1, OFD1, OPHN1, OTUD6B, PACS2, PAK1, PAK3, PARS2, PCDH19, PGAP1, PHACTR1, PHF6, PHGDH, PIGA, PIGB, PIGG, PIGL, PIGN, PIGO, PIGP, PIGT, PIGU, PIGW, PLCB1, PLP1, PLPBP, PNKP, PNPO, POLG, PPP2CA, PPP3CA, PPT1, PQBP1, PRICKLE1, PRIMA1, PRRT2, PSAT1, PSPH, PURA, QARS1, RAB39B, RAI1, RANBP2, RELN, RHOBTB2, RNASEH2A, RNASEH2B, RNASEH2C, ROGDI, RORA, RORB, RPS6KA3, SAMHD1, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SETD1A, SHANK3, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC1A2, SLC1A3, SLC1A4, SLC25A1, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SLC6A5, SLC6A8, SLC9A6, SMC1A, SMS, SNAP25, SON, SPATA5, SPTAN1, ST3GAL3, ST3GAL5, STAMBP, STRADA, STX1B, STXB1, SYN1, SYNGAP1, SYNJ1, SYP, SZT2, TANC2, TANGO2, TBC1D24, TBCE, TBCK, TCF4, TDP2, TPP1, TRAK1, TREX1, TRIM8, TRIO, TRPM6, TSC1, TSC2, UBA5, UBE2A, UBE3A, UFM1, UGDH, UGP2, WDR45, WWOX, YWHAG, ZDHHC9, ZEB2

Repeatexpansieanalyse: CSTB

Benign neonatal/infantile convulsions (EPI01v21.1; 7 genes)
Including deletion/duplication test for KCNQ2

ATP1A1, KCNQ2, KCNQ3, PRRT2, SCN2A, SCN8A, TBC1D24

Focal epilepsy (EPI04v21.1; 22 genes)

CHRNA2, CHRNA4, CHRN2, CNKSR2, DCX, DEPDC5, FLNA, GRIN2A, KCNT1, LGI1, MICAL1, MTOR, NPRL2, NPRL3, POLG, PRIMA1, RELN, SLC12A5, SYN1, TSC1, TSC2, ZDHHC9

Epilepsy (febrile/inflammatory, generalized and/or paroxysmal) (EPI11v21.1; 29 genes)

ATP1A2, ATP1A3, CACNA1A, CHD2, CLCN4, CPT2, CSTB, GABRA1, GABRB3, GABRG2, HCN1, HCN2, KCNA1, KCNA2, KCNM1A, MAST3, PCDH19, POLG, PRRT2, RANBP2, RORB, SCN1A, SCN1B, SCN2A, SCN8A, SLC1A3, SLC2A1, STX1B, TBC1D24

Deletion/duplication test: SCN1A SLC2A1 PCDH19 CSTB

In submitting this sample the clinician confirms that the patient has been informed about the chances of uncovering incidental findings that can result from this medical test.

[§] Sequence and copy number analysis

[^] Repeat expansion analysis only

Epilepsy

Individual genes | Sequence analysis

- Dravet syndrome (SMEI/SMEB)[§] SCN1A[§]
 - Progressive myoclonic epilepsy type 1 / Unverricht Lundborg Disease (ULD) CSTB
- Including repeatexpansion test

Hereditary cancer

Gene panels (incl. WES based CNV analysis per gene)

- Breast - and ovarium cancer** (ONC02v22.1; 10 genes)
BRCA1 copynumber analysis included
BRCA1, BRCA2, ATM, BARD1, CHEK2, PALB2, RAD51C, RAD51D, PTEN, BRIP1
- Ovarian cancer** (ONC01v22.1; 6 genes)
BRCA1 copynumber analysis included
BRCA1 , BRCA2*, PALB2*, RAD51C, RAD51D, BRIP1, RAD51D*
- Pheochromocytoma** (ONC04v18.1; 11 genes)
SDHAF2 and VHL copy number analysis included.
FH, MAX, MDH2, RET (relevant exons only), SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL
- Paraganglioma** (ONC05v18.1; 6 genes)
SDHAF2 copy number analysis included.
MAX, SDHA, SDHAF2, SDHB, SDHC, SDHD
- Pancreatic cancer** (ONC13v22.1; 6 genes)
BRCA1 copy number analysis included.
ATM, BRCA1 , BRCA2, CDKN2A, PALB2, STK11
- MEN related disorders** (ONC06v23.1; 11 genes)
Copy number analysis included for: AIP, CDKN1B en MEN1.
AIP, AP2S1, CASR, CDC73, CDKN1A, CDKN1B, CDKN2B, CDKN2C, GNA11, MEN1, RET
- Renal cancer** (ONC07v18.1; 7 genes)
VHL copy number analysis included.
BAP1, FH, FLCN, MET, PTEN, SDHB, VHL
- Wilms tumor predisposition** (ONC03v23.1; 29 genes + 1 microdeletion region)
AMER1, ASXL1, BLM, BRCA2, BUB1B, CDC73, CDKN1C, CEP57, CTR9, DICER1, DIS3L2, FBXW7, GPC3, GPC4, HACE1, MLH1, MSH2, MSH6, NF1, NYNRIN, PALB2, PIK3CA, PMS2, REST, TP53, TRIM28, TRIM37, TRIP13, WT1, 9q22.3 microdeletion region
- Polyposis/colorectal cancer** (ONC08v20.1; 19 genes)
APC, MUTYH (6 out of 16 exons). copy number analysis included.
APC, BMPR1A, EPCAM, GREM1, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2 (reduced sensitivity due to pseudogene presence), POLD1, POLE, PTEN, RNF43, RPS20, SMAD4, STK11
- Non-polyposis/colorectal cancer** (ONC09v20.1; 7 genes)
MSH6, MLH1 and MSH2 copy number analysis included.
EPCAM, MLH1, MSH2, MSH6, PMS2 (reduced sensitivity due to pseudogene presence), POLD1, POLE
- Prostate cancer** (ONC11v21.1; 5 genes)
BRCA1 copy number analysis included
BRCA1, BRCA2, ATM, CHEK2, PALB2
- Pediatric cancer predisposition** (ONC14v23.1; 140 genes)
NB: Requests for this panel are reserved exclusively for clinical geneticists
ABCB11, ACD, AIP, ALK, AMER1, APC, ATM, BAP1, BLM, BRAF, BRCA2, BRIP1, BUB1B, CBL, CD27, CD70, CDC73, CDH1, CDKN1C, CDKN2A, CE2BP1, CEP57, CREBBP, CTC1, CTLL4, CTR9, DDB2, DICER1, DIS3L2,

DKC1, EGLN1, EGLN2, EPAS1, EPCAM, ERCC2, ERCC3, ERCC4, ERCC5, ETV6, EZH2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FAS, FBXW7, FH, GATA2, GPC3, GPR161, HAVCR2, HRAS, IKBKAP, IKZF1, ITK, KRAS, LIG4, LZTR1, MAP2K1.....
...MAP2K2, MDH2, MEN1, MLH1, MSH2, MSH6, NBN, NF1, NF2, NHP2, NOP10, NRAS, NSD1, PALB2, PARN, PAX5, PHOX2B, PIK3CA, PMS2, POLD1, POLE, POLH, PTCH1, PTEN, PTPN11, RAF1, RB1, RECQL4, REST, RET, RIT1, RPL11, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS27, RRAS, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SETBP1, SH2D1A, SHOC2, SMARCA4, SMARCB1, SMARCE1, SOS1, STK11, SUFU, TCF3, TERT, TIN2, TP53, TRIM28, TRIM37, TRIP13, TSC1, TSC2, TYK2, USB1, VHL, WAS, WRAP53, WT1, XPA, XPC

Hereditary cancer

Single gene | Sequence analysis

- Risc factor for o.a. breastcancer / Ataxia-telangiectasia ATM
- Risc factor for o.a. breastcancer (CHEK2) CHEK2
- PTEN Hamartoma tumor syndrome (PHTS) PTEN
- Lynch syndrome (HNPCC2)[§] MLH1[§]
- Lynch syndrome (HNPCC1)[§] MSH2[§]
- Lynch syndrome (HNPCC5)[§] MSH6[§]
- Multiple Endocrine Neoplasia type 1 (MEN1)[§] MEN1[§]
- Multiple Endocrine Neoplasia type 2 (MEN2) RET
(only relevant exons)
- Medullary thyroic cancer, sporadic (SMTC) RET
(on tumor tissue derived DNA only)
- Von Hippel-Lindau, disease (VHL)[§] VHL[§]

Intellectual disability: syndromal/non-syndromal

Gene panel | Exome (incl. WES based CNV analysis per gene)

This gene panel, and the exome-wide analysis, can only be requested by clinical geneticists. Contact us for more information.

Exome trio analysis intellectual disability

(VBE01v23.1; 1601 genes/exome)

This gene panel includes a CNV analysis for known micro deletion & duplication syndromes (for a specification of the regions, see link below)

For an overview of the genes included in the gene panel see:

<http://www.umcutrecht.nl/nl/Ziekenhuis/Professionals/Diagnostiek-aanvragen/Genoomdiagnostiek/Next-Generation-Sequencing-NGS>

Intellectual disability: syndromal/non-syndromal

Single gene | Sequence analysis

- Angelman syndrome (AS) (*methylation-sensitive deletion/duplication test*) [15q11-q13]
- Cohen syndrome[§] VPS13B[§]
- Fragile-X syndrome, FRAXA ^ FMR1^
- Lesch-Nyhan syndrome, (LNS) HPRT1
- Rett syndrome, RTT[§] MECP2[§]
- Rett syndrome, atypical[§] CDKL5[§]
- Rett syndrome, congenital variant[§] FOXG1[§]
- Prader-Willi syndrome (PWS) (*methylation-sensitive deletion/duplication test*) [15q11-q13]

In submitting this sample the clinician confirms that the patient has been informed about the chances of uncovering incidental findings that can result from this medical test.

[§] Sequence and copy number analysis

^ Repeat expansion analysis only

Liver diseases

Gene panels (incl. WES based CNV analysis per gene)

- Intrahepatic cholestasis** (HEP01v24.1 (formerly known as MET02); 10 genes)
ABCB11, ABCB4, ATP8B1, KIF12, LSR, MYO5B, NR1H4, TJP2, USP53, ZFYVE19
- Cholestasis, broad differential diagnosis** (HEP02v24.1 (formerly known as MET10); 81 genes)
ABCB11, ABCB4, ABCC2, ABCD3, ADK, AHCY, AKR1D1, ALDOB, AMACR, ARG1, ASAH1, ATP7B, ATP8B1, BAAT, BCS1L, C10orf2, CFTR, CIRH1A, CLDN1, CYP27A1, CYP7B1, DCDC2, DGUOK, DHCR7, FAH, GALT, GBA, GBE1, GLIS3, HADHA, HNF1A, HNF1B, HSD3B7, IFT43, INVS, JAG1, KIF12, LIPA, LSR, MPV17, MTM1, MYO5B, NOTCH2, NPC1, NPC2, NPHP3, NR1H4, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX6, POLG, POMC, PROP1, SCO1, SERPINA1, SHPK, SLC25A13, SLC27A5, SLC01B1, SLC01B3, STX3, SUCLA2, TALDO1, TJP2, TPO, TRMU, TULP3, UGT1A1, UNC45A, USP53, VIPAS39, VPS33B, ZFYVE19

Metabolic diseases

Gene panels (incl. WES based CNV analysis per gene)

Please note: gene panels 'Intrahepatic cholestasis (MET02)' & 'Cholestasis, broad differential diagnosis (MET10)' are now available under 'Liver diseases'.

- Glycin encephalopathy / non-ketonic hyperglycinemia** (MET01v22.1; 3 genes)
AMT, GCSH, GLDC
- Glycogen storage disease** (MET06v16.2; 23 genes)
AGL, ENO3, GAA, GBE1, GYG1, GYS1, LDHA, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PYGL, PYGM, SLC2A2, G6PC, PHKG2, PHKB, ALDOA, GYS2, SLC37A4, LAMP2, PRKAG2
- Serine synthesis defect** (MET03v16.1; 3 genes)
PHGDH, PSPH, PSAT1
- Niemann-Pick disease** (MET04v16.1; 3 genes)
SMPD1, NPC1, NPC2
- Methylmalonic aciduria (MMA)** (MET11v20.1; 29 genes)
ABCD4, ACSF3, ALDH6A1, AMN, CBS, CD320, CLYBL, CUBN, GIF, HCFC1, HIBCH, IVD, LMBRD1, MCEE, MMAA, MMAB, MMACHC, MMADHC, MTHFR, MTR, MTRR, MUT, SLC46A1, SUCLA2, SUCLG1, TCN1, TCN2, THAP11, ZNF143

Metabolic diseases

Single gene | Sequence analysis

- Biotinidase deficiency BTD
- Congenital disorder of glycosylation type 1A (CDG1A) PMM2
- Congenital disorder of glycosylation type 1P (CDG1P) ALG11
- Congenital disorder of glycosylation type 3 (CDG3) COG6
- Familial Hyper insulinic Hypoglycemia type 7, (HHF7) SLC16A1
- Phenylketonuria type 1 (PKU) PAH
- Phenylketonuria type 3 (PTPS) PTS
- Glycerol kinase deficiency (GKD) GK
- Hartnup disorder SLC6A19
- Hemochromatosis, (HFE) HFE
- Medium-Chain Acyl-CoA Dehydrogenase deficiency (MCAD) ACADM
- Metachromatic Leukodystrofia (MLD) ARSA
- Pompe, Disease, Glycogen storage disease II (GSD2)[§], including deletion test exon 18 GAA[§]
- Pyruvaat Kinase deficiency (PK) PKLR
- Tyrosinemia, type I FAH
- Wilson disease (WD) ATP7B

In submitting this sample the clinician confirms that the patient has been informed about the chances of uncovering incidental findings that can result from this medical test.

[§] Sequence and copy number analysis

Neurological disorders

Gene panels (incl. WES based CNV analysis per gene)

• Repeat expansions are found to underlie a substantial part of neuromuscular diseases. These cannot be detected using NGS sequencing and should be requested separately by checking the boxes.

- FTD-ALS*** (NEU01v22.1; 28 genes)
Includes repeat expansion analysis SCA2 (ATXN2)
ALS2, ANG, ANXA11, APP, C21ORF2, CHCHD10, CHMP2B, ERBB4, FUS, GRN, KIF5A, MAPT, MATR3, NEK1, OPTN, PFN1, PRPH, PSEN1, PSEN2, SETX, SIGMAR1, SOD1, TARDBP, TBK1, TUBA4A, UBQLN2, VAPB, VCP
Repeat expansion analysis*: C9ORF72
- Cerebral cavernous malformations (CCM)** (NEU03v16.1; 3 genes)
Includes copy number analysis of KRIT1
KRIT1, CCM2, PDCD10
- Fahr disease** (NEU04v24.1; 8 genes)
JAM2, KIAA1161, NAA60, PDGFB, PDGFRB, SLC20A2, XPR1, CMPK2
- Sporadic ALS** (NEU05v22.1; 2 genes)
FUS, SOD1
Includes repeat expansion analysis for C9ORF72 and SCA2 genes

Neurological disorders

Single gene | Sequence | repeat analysis

- Amyotrofe lateral sclerosis / Frontotemporal dementia (ALS/FTD)[^] C9ORF72[^]
- SCA2 (Riskfactor for ALS)[^] ATXN2[^]

Neuromuscular disease

Gene panels (incl. WES based CNV analysis per gene)

As of Januari 1st 2024 we do not perform genetic diagnostic testing for ataxia. Please consult the UMC Groningen (UMCG expertcentre movement disorders) and/or RadboudUMC (expertcentre rare and hereditary movement disorders RadboudUMC) regarding genetic diagnostic testing for ataxia.

• Repeat expansions are found to underlie a substantial part of neuromuscular diseases. These cannot be detected using NGS sequencing and should be requested separately by checking the boxes.

- Broad NMD panel** (NEM27v22.1; 430 genes)
AARS1, ABCD1, ABHD5, ACAD9, ACADVL, ACTA1, ACTN2, ACVR1, COQ8A, ADSS1, AFG3L2, AGL, AGRN, AIFM1, ALDH18A1, ALDH3A2, ALG13, ALG14, ALG2, ALS2, AMPD2, ANO10, ANO5, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APTX, AR, ARHGEF10, ARL6IP1, ARSA, ASAH1, ASCC1, ATG7, ATL1, ATL3, ATM, ATP13A2, ATP1A1, ATP2A1, ATP7A, B3GALNT2, B4GALNT1, B4GAT1, BAG3, BEAN1, BICD2, BIN1, BSCL2, BVES, C12orf65, C19orf12, CACNA1A, CACNA1G, CACNA1S, CACNB4, CAPN1, CAPN3, CASQ1, CAV3, CCDC78, CCDC88C, CCT5, CFL2, CHAT, CHCHD10, CHKB, CHRNA1, CHRN1B, CHRNA2, CHRNE, CHRNG, CLCN1, CLN3, CNTN1, CNTNAP1, COA7, COL12A1, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, COX6A1, CPT2, CRYAB, CTDP1, CWF19L1, CYP21A1, CYP7B1, DAG1, DCAF8, DCTN1, DDHD1, DDHD2, DES, DGAT2, DHTKD1, DMD, DNAJB2, DNAJB6, DNMT2, DNMT1, DOK7, DPAGT1, DPM1, DPM2, DPM3, DST, DYNCH1H1, DYSF, EBF3, ECEL1, EEF2, EGR2, ELOVL4, ELOVL5, ELP1, EMD, ENO3, ENTDP1, ERBB3, ERLIN1, ERLIN2, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, FA2H, FAM111B, RETREG1, FARS2, FASTKD2, FBLS, FBXO38, FGD4, FGF14, FHL1, FIG4, FKBP, FKTN, FLAD1, FLNC, FXN, FXR1, GAA, GAN, GARS1, GBA2, GBE1, GDAP1, GDAP2, GFPT1, GJB1, GJB3, GJC2, GLA, GLE1, GMPPB, GNB4, GNE, GOLGA2, GRID2, GRM1, GYG1, GYS1, HACE1, HARS1, HEXB, HINT1, HK1, HNRNPA1, HNRNPDL, HNRNPA2B1, HOXD10, HRAS, HSPB1, HSPB3, HSPB8, HSPD1, HSPG2, IBA57, IFRD1, IGHMBP2, INF2, INPP5K, ISCU, CRPPA, ITGA7, ITPR1, KARS1, KBTBD13, KCNA1, KCNC3, KCND3, KCNE1, KCNE5, KCNE2, KCNE3, KCNJ18, KCNJ2, KCNJ5, WASHC5, KIDINS220, KIF1A, KIF1B, KIF1C, KIF21A, KIF5A, KLC2, KLHL40, KLHL41, KLHL9, KY, LITCAM, LAMA2, LAMB2, LAMP2, LARG1, LDB3, LDHA, LIMS2, LITAF, LMNA, LMOD3, LPIN1, LRP4, LRSAM1, MAG, MAP3K20, MARS1, MARS2, MED25, MEGF10, MFN2, MICU1, MME, MORC2, MPZ, MRE11, MSTN, MSTO1, MTM1, MTMR2, MTPAP, MUSK, MYBP3, MYF6, MYH2, MYH3, MYH7, MYH8, MYL1, MYL2, MYMK, MYO18B, MYO9A, MYOT, MYPN, NAGLU, NDRG1...

[^] Repeat expansion analysis only

Neuromuscular disease (continued)

Gene panels (incl. WES based CNV analysis per gene)

... NEB, NEFH, NEFL, NGF, NIPA1, NOP56, NTSC2, NTRK1, OBSCN, OPA1, ORA1, PABPN1, PAX7, PDK3, PODYN, PEXT, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHOX2A, PHYH, PIP5K1C, PLEC, PLEKHG5, PLP1, PMP2, PMP22, PNKP, PNPLA2, PNPLA6, PNPLA8, POGUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, PRDM12, PREPL, PRKAG2, PRKCG, PRPS1, PRX, HACD1, CAVIN1, PTRH2, PUS1, PYGM, PYROXD1, RAB7A, RAPSIN, RBCK1, REEP1, RNF216, RRM2B, RTN2, RXYLT1, RYR1, SACS, SBF1, SBF2, SCN10A, SCN11A, SCN4A, SCN9A, SCYL1, SELENON, SEPTIN9, SETX, SGCA, SGCB, SGCD, SGCE, SGGC, SGPL1, SH3TC2, SIGMAR1, SIL1, SLC12A6, SLC1A3, SLC22A5, SLC25A20, SLC25A4, SLC25A42, SLC33A1, SLC52A2, SLC52A3, SLC5A7, SMCHD1, SNAP25, SINX14, SORD, SPAST, SPEG, SPG11, SPART, SPG21, SPG7, SPTBN2, SPTBN4, SPTLC1, SPTLC2, SQSTM1, STAC3, STIM1, STUB1, SUCLA2, SURF1, SYNE1, SYNE2, SYT2, TCAP, TDP1, TDP2, TECPR2, TFG, TGM6, THG1L, TIA1, TK2, TMEM240, TMEM43, TMEM65, TNNI2, TNNT1, TNNT3, TNPO3, TOR1A, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM2, TRIM32, TRIM54, TRIM63, TRIP4, TRPC3, TRPV4, TTBK2, TTC19, TTN, TTPA, TTR, TUBB3, TWNK, UBA1, UBAP1, VAMP1, VCP, VIPAS39, VMA21, VPS13D, VPS37A, VRK1, WARS1, WNK1, YARS1, YARS2, ZFYVE26, ZFYVE27

Congenital/metabolic myasthenic syndromes (NEM12v22.1; 29 genes)
 AGRN, ALG14, ALG2, CHAT, CHRNA1, CHRN1B, CHRND, CHRNE, CHRNG, COL13A1, COLQ, DOK7, DPAGT1, GFPT1, GMPPB, LAMB2, LRP4, MUSK, MYO9A, PLEC, PREPL, RAPSIN, SCN4A, SLC18A3, SLC25A1, SLC5A7, SNAP25, SYT2, VAMP1

Congenital muscular dystrophy (NEM07v19.1; 34 genes)
 ACTA1, ALG13, B3GALNT2, B4GAT1, CHKB, COL12A1, COL6A1, COL6A2, COL6A3, DAG1, DNM2, DPM1, DPM2, FHL1, FKRP, FKTN, GMPPB, GOLGA2, INPP5K, ISPD, ITGA7, LAMA2, LARGE1, LMNA, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1, SELENON, TCAP, TRAPPC11, TRIP4

Congenital myopathy (NEM04v22.1; 39 genes)
 ACTA1, ACTN2, BIN1, CACNA1S, CFL2, CNTN1, DNM2, FXR1, HACD1, HNRNPA1, HRAS, KBTBD13, KLHL40, KLHL41, LMOD3, MAP3K20, MEGF10, MTM1, MYBPC3, MYH2, MYH7, MYL1, MYL2, MYMK, MYO18B, MYPN, NEB, PAX7, PYROXD1, RYR1, SELENON, SPEG, SPTBN4, STAC3, TNNT1, TPM2, TPM3, TRIM32, TTN

Distal myopathy (NEM05v22.1; 24 genes)
 ACTN2, ADSS1, ANO5, BAG3, CAV3, CRYAB, DES, DNM2, DYSF, FLNC, GNE, KLHL9, KY, LDB3, MATR3, MYH7, MYOT, NEB, SELENON, SORD, SQSTM1, TIA1, TTN, VCP

Hereditary spastic paraplegia (HSP) (NEM26v22.1; 61 genes)
 ABCD1, AFG3L2, ALDH18A1, ALDH3A2, ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BSCL2, C12orf65, C19orf12, CAPN1, CYP2U1, CYP7B1, DDHD1, DDHD2, ENTDP1, ERLIN1, ERLIN2, FA2H, FARS2, GBA2, GJC2, HACE1, HSPD1, IBA57, WASHC5, KIDINS220, KIF1A, KIF1C, KIF5A, KLC2, L1CAM, MAG, MARS2, MTPAP, NIPA1, NTSC2, PLP1, PNPLA6, REEP1, RTN2, SACS, SLC33A1, SPAST, SPG11, SPART, SPG21, SPG7, TECPR2, TFG, UBAP1, VAMP1, VPS37A, ZFYVE26, ZFYVE27

Limb-Girdle muscle weakness (NEM08v22.1; 44 genes)
 ANO5, BVES, CAPN3, CAV3, DAG1, DES, DMD, DNAJB6, DPM3, DYSF, EMD, FHL1, FKRP, FKTN, GAA, GMPPB, HNRNPDL, ISPD, LIMS2, LMNA, MYOT, PLEC, POGUT1, POMGNT1, POMT1, POMT2, POPDC3, PTRF, PYROXD1, SGCA, SGCB, SGCD, SGGC, SMCHD1, SYNE1, SYNE2, TCAP, TMEM43, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN, VCP

Malignant hyperthermia (NEM11v17.1; 3 genes)
 CACNA1S, RYR1, SCN4A

Metabolic myopathy (NEM30v19.1; 28 genes)
 ABHD5, ACAD9, ACADVL, AGL, CPT2, ENO3, ETFA, ETFB, ETFDH, FLAD1, GAA, GBE1, GYG1, GYS1, LDHA, LPIN1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PNPLA2, PNPLA8, PRKAG2, PYGM, RBCK1, SLC22A5, SLC25A20

Motor neuron disease (MND) (NEM13v22.1; 56 genes)
 AARS1, ALS2, ANG, ANXA11, AR, ASAH1, ASCC1, ATP7A, BICD2, BSCL2, CHCHD10, CHMP2B, DCTN1, DNAJB2, DYNC1H1, ERBB3, ERBB4, EXOSC3, EXOSC8, FBXO38, FIG4, FUS, GARS1, GLE1, HEXB, HNRNPA1, HNRNPA2B1, HSPB1, HSPB3, IGHMBP2, MATR3, NEFH, OPTN, PFN1, PIP5K1C, PLEKHG5, PRPH, REEP1, SETX, SIGMAR1, SLC52A2, SLC52A3, SLC5A7, SOD1, SPG11, SQSTM1, TARDBP, TRIP4, TRPV4, TUBA4A, UBA1, UBQLN2, VAPB, VCP, VRK1, WARS1

Repeat expansion analysis[•]: C9ORF72
Copy number analysis: SMN1/(SMN2)

Motor and Sensory Neuropathy (NEM15v22.1; 91 genes)
 AARS1, AIFM1, ARHGEF10, ARSA, ATL1, ATL3, ATP1A1, BAG3, BSCL2, CCT5, COX6A1, CTDP1, DCAF8, DGAT2, DHTKD1, DNAJB2, DNM2, DNMT1, DST, DYNC1H1, EGR2, ELP1, RETREG1, FBLN5, FGD4, FIG4,

GAN, GARS1, GDAP1, GJB1, GJB3, GLA, GNB4, HARS1, HINT1, HK1, HOXD10, HSPB1, HSPB3, HSPB8, IGHMBP2, INF2, KARS1, KIF1A, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, MARS1, MFN2, MME, MORC2, MPZ, MTMR2, NAGLU, NDRG1, NEFH, NEFL, NGF, NTRK1, PDK3, PLEKHG5, PMP2, PMP22, PNKP, PRDM12, PRPS1, PRX, RAB7A, SBF1, SBF2, SCN10A, SCN11A, SCN9A, SEPTIN9, SH3TC2, SLC12A6, SORD, SPG11, SPTLC1, SPTLC2, SURF1, TFG, TRIM2, TRPV4, TTR, VCP, VRK1, WNK1, YARS1

Myotonic syndromes (NEM09v22.1; 5 genes)
 ATP2A1, CAV3, CLCN1, HSPG2, SCN4A

Repeat expansion analysis[•]: DMPK CNBP

NMDs with episodic attacks (NEM28v22.1; 15 genes)
 CACNA1A, CACNA1S, CLCN1, KCNA1, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ18, KCNJ2, KCNQ1, OBSCN, RYR1, SCN4A, SCN5A

Periodic paralysis and ion channel muscle disease (NEM10v22.1; 12 genes)
 CACNA1A, CACNA1S, CLCN1, KCNA1, KCNE1, KCNE5, KCNE2, KCNE3, KCNJ5, KCNJ18, KCNJ2, SCN4A

Scapulo-peroneal syndromes (NEM25v16.1; 13 genes)
 CAPN3, DES, EMD, FHL1, GAA, LAMP2, LMNA, MYH7, PYGM, SYNE1, SYNE2, TMEM43, TRPV4

Other neuromuscular disease (NEM20v19.1; 34 genes)
 AIFM1, CASQ1, CHCHD10, CNTNAP1, ELP1, FAM111B, FASTKD2, KIF21A, MYH3, MYH8, OPA1, ORA1, PHOX2A, POLG, POLG2, PTRH2, PUS1, RRM2B, SGCE, SLC25A4, SLC25A42, STIM1, SUCLA2, SYNE1, TK2, TMEM65, TNNI2, TNNT3, TOR1A, TPM2, TTR, TUBB3, TWNK, YARS2

Neuromuscular diseases

Single gene | Sequence analysis

- Central core disease/malignant hyperthermia RYR1
- Kennedy disease: X-bound type 1 SBMA, (SMAX1)[^] AR[^]
- Motor en sensory neuropathy PMP22/MPZ/GJB1
(deletion/duplication test only)
- Myotonic dystrophy type 1 (DM1)[^] DMPK[^]
- Myotonic dystrophy type 2 (DM2)[^] CNBP[^]
- Spinal Muscular Atrophy (SMA type 1 - 4)[§] (sequence-analysis only after consultation) SMN1[§]

Obesity

Gene panels (incl. WES based CNV analysis per gene)

Obesity (OBE02v22.1, 5 genes)
 LEP, LEPR, PCSK1, POMC, MC4R

Obesity

Single gene | Sequence analysis

Cohen syndrome[§] VPS13B[§]

In submitting this sample the clinician confirms that the patient has been informed about the chances of uncovering incidental findings that can result from this medical test.

[§] Sequence and copy number analysis

[^] Repeat expansion analysis only

Primary immunodeficiencies

Gene panels (incl. WES based CNV analysis per gene)

- ALPS/Autoimmunity (PID03v17.1; 12 genes)**
FAS, FASLG, CASP10, CASP8, KRAS, NRAS, FADD, AIRE, FOXP3, IL2RA, ITCH, LRBA
- Autoinflammatory disease (PID01v22.2; 67 genes)**
ACP5, ADA2, ADAM17, ADAR, ADGRE2, ALPK1, AP1S3, C2orf69, CARD14, CDC42, CEBPE, COPA, DDX58, DNASE1, DNASE1L3, DNASE2, FERMT1, IFIH1, IKZF1, IL10, IL10RA, IL10RB, IL1RN, IL36RN, LACC1, LPIN2, LSM11, MEFV, MVK, NCKAP1L, NCSTN, NLR4, NLRP1, NLRP2, NLRP3, NOD2, OTULIN, PEPD, PIK3CD, PLCG2, POMP, PRKCD, PSENEN, PSMA3, PSMB4, PSMB8, PSMB9, PSMG2, PSTPIP1, RBCK1, RIPK1, RNASEH2A, RNASEH2B, RNASEH2C, RNF31, SAMHD1, SLC29A3, STAT2, STING1, SYK, TNFAIP3, TNFRSF1A, TREX1, TRNT1, UBA1, USP18, WDR1
- Autoinflammatory mosaicism (PID09v24.1; 6 genes)**
Analysis of mosaic variants in the following genes:
NLR4, NLRP3, NOD2, PSTPIP1, TNFRSF1A, UBA1
- B-cell pathology (PID05v16.1; 14 genes)**
BTK, ICOS, CD19, CD81, TNFRSF13B, TNFRSF13C, CD40, CD40L, AICDA, UNG, CD79A, BLNK, CD79B, IGLL1
- Chronic mucocutaneous candidiasis (CMC) (PID07v17.1; 7 genes)**
IL17RA, IL17F, STAT1, TLR3, AIRE, IL2RA, CARD9
- HLH/Immune dysregulation (PID02v22.1; 21 genes)**
AP1S3, AP3B1, AP3D1, CD27, CD70, CORO1A, CTPS1, FAAP24, ITK, LYST, MAGT1, PRF1, RAB27A, RASGRP1, RC3H1, RHOG, SH2D1A, STX11, STXB2P, UNC13D, XIAP
Copy number analysis: PRF1 UNC13D STX11
- Hyper IgE Syndromes (HIES) (PID06v21.1; 9 genes)**
CARD11, CARD14, DOCK8, IL6R, IL6ST, PGM3, STAT3, TYK2, ZNF341
- (S)CID (PID04v20.1; 29 genes)**
ADA, AK2, BCL11B, CD3D, CD3E, CD3G, CD40, CD40LG, CD8A, CORO1A, DCLRE1C, DOCK8, FOXN1, IL2RA, IL2RG, IL7R, JAK3, LIG4, NHEJ1, ORAI1, PNP, PRKDC, PTPRC, RAG1, RAG2, STAT5B, STIM1, TBX1, ZAP70
- Primary immunodeficiencies full panel (PID00v23.1; 468 genes)**

The following non-coding RNA genes are not included in this analysis: *RMRP, RNU4ATAC, RNU7-1, SNORA31 and TERC*. Please contact us for diagnostic opportunities if there's a strong suspicion of involvement for these non-coding genes.

ACD, ACP5, ACTB, ADA, ADA2, ADAM17, ADAR, AGA, AICDA, AIRE, AK2, ALG13, ALPI, ALPK1, ANGPT1, AP1S3, AP3B1, AP3D1, APOL1, ARHGEF1, ARPC1B, ATAD3A, ATG4A, ATM, ATP6AP1, B2M, BACH2, BCL10, BCL11B, BLK, BLM, BLNK, BLOC1S6, BTK, C1QA, C1QB, C1QC, C1R, C1S, C2, C2orf69, C3, C5, C6, C7, C8A, C8B, C8G, C9, CA2, CARD10, CARD11, CARD14, CARD9, CARMIL2, CASP10, CASP8, CAVIN1, CCBE1, CD19, CD247, CD27, CD28, CD3D, CD3E, CD3G, CD4, CD40, CD40LG, CD46, CD55, CD59, CD70, CD79A, CD79B, CD81, CD8A, CDC42, CDCA7, CDKN2B, CEBPE, CFB, CFD, CFH, CFI, CFP, CFTR, CHD7, CHUK, CIB1, CIITA, CLCN7, CLEC4D, CLEC7A, CLPB, COPA, COPG1, CORO1A, CR2, CRACR2A, CREBBP, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTNND1, CTPS1, CTSC, CXCR2, CXCR4, CYBA, CYBB, CYBC1, DBR1, DCLRE1B, DCLRE1C, DDX58, DEF6, DGAT1, DHFR, DIAPH1, DKC1, DNAJC21, DNASE1, DNASE1L3, DNASE2, DNMT3B, DOCK2, DOCK8, ELANE, ELF4, EPG5, ERBIN, ERCC2, ERCC3, ERCC6L2, EXTL3, F12, FAAP24, FADD, FAS, FASLG, FAT4, FCGR3A, FCHO1, FCN3, FERMT1, FERMT3, FNIP1, FOXI3, FOXN1, FOXP3, FPR1, G6PC, G6PC3, G6PD, GATA2, GF11, GINS1, GJC2, GRHL2, GTF2H5, HAVCR2, HAX1, HCK, HELLS, HMOX1, HS3ST6, HYOU1, ICOS, ICOSLG, IFIH1, IFNAR1, IFNAR2, IFNG, IFNGR1, IFNGR2, IGHM, IGLL1, IKKBK, IKKKG, IKZF1, IKZF2, IKZF3, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL17F, IL17RA, IL17RC, IL18BP, IL1RN, IL2, IL21, IL21R, IL2RA, IL2RB, IL2RG, IL36RN, IL6R, IL6ST, IL7R, INO80, INSR, IRAK1, IRAK4, IRF2BP2, IRF3, IRF4, IRF7, IRF8, IRF9, ISG15, ITCH, ITGB2, ITK, ITPKB, ITRP3, IVNS1ABP, JAGN1, JAK1, JAK2, JAK3, KDM6A, KMT2A, KMT2D, KNG1, KRAS, LACC1, LAMTOR2, LAT, LCK, LCP2, LIG1, LIG4, LPIN2, LRBA, LRRC8A, LSM11, LYST, MAGT1, MALT1, MAN2B1, MAN2B2, MANBA, MAP1LC3B2, MAP3K14, MAPK8, MASP2, MC2R, MCM10, MCM4, MEFV, MOGS, MPEG1, MRFA, MS4A1, MSN, MTHFD1, MVK, MYD88, MYOF, MYSM1, NBAS, NBN, NCF1, NCF2, NCF4, NCKAP1L, NCSTN, NFAT5, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLR4, NLRP1, NLRP2, NLRP3, NOD2, NOD1, NOS2, NRAS, NSMCE3, OAS1, ORAI1, OSTM1, OTULIN, PARN, PAX1, PAX5, PBX1, PCCA, PCCB, PDCD1, PEPD, PGM3, PIGA, PIK3CD, PIK3CG, PIK3R1, PLCG2, PLEKHM1, PLG, PNM2, PNP, POLA1, POLE2, POLR3F, POMP, POT1, POU2AF1, PRF1, PRKCD, PRKDC, PRPS1, PSENEN, PSMA3, PSMB4, PSMB8, PSMB9, PSMG2, PSTPIP1, PTPN22, PTPRC, RAB27A, RAC2, RAG1, RAG2, RANBP2, ...

...RASGRP1, RASGRP2, RBCK1, RC3H1, RECQL4, REL, RELA, RELB, RFX5, RFXANK, RFXAP, RHOG, RHOH, RIPK1, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RORC, RPA1, RPSA, RSPH9, RTEL1, SAMD9, SAMD9L, SAMHD1, SASH3, SAT1, SBD5, SEC61A1, SEMA3E, SERAC1, SERPING1, SH2B3, SH2D1A, SH3BP2, SH3KBP1, SKIV2L, SLC29A3, SLC35A1, SLC35C1, SLC37A4, SLC39A4, SLC39A7, SLC46A1, SLC7A7, SMARCAL1, SMARCD2, SNX10, SOCS1, SOCS4, SP110, SPI1, SPINK5, SPPL2A, SRP72, STAT1, STAT2, STAT3, STAT4, STAT5B, STAT6, STIM1, STING1, STK4, STN1, STX11, STXB2P, SYK, TAP1, TAP2, TAPBP, TAZ, TBX1, TBX21, TCF3, TCIRG1, TGN2, TERT, TET2, TFRC, TGFBI, THBD, TICAM1, TINF2, TIRAP, TLR3, TLR4, TLR7, TLR8, TMC6, TMC8, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF14, TNFRSF4, TNFRSF9, TNFSF11, TNFSF12, TNFSF13, TOM1, TOP2B, TPP2, TRAC, TRAF3, TRAF3IP2, TREX1, TRIM22, TRNT1, TTC37, TTC7A, TYK2, UBA1, UNC13D, UNC93B1, UNG, USB1, USP18, VAV1, VPS13B, VPS45, WAS, WDR1, WIPF1, WRAP53, XIAP, ZAP70, ZBTB24, ZNF341, ZNF41

Primary immunodeficiencies

Single gene | Sequence analysis

- VEXAS syndrome UBA1

Renal disease

Gene panels (incl. WES based CNV analysis per gene)

See Hereditary cancer for the renal cancer panel.

- Atypical Hemolytic uremic syndrome (aHUS)/ Thrombotic microangiopathies (NEF07v23.1; 15 genes)**
Includes copy number analysis of CD46, CFH, CFI, CFHR1, CFHR3
ADAMTS13, C1GALT1C1, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFI, DGKE, MMACHC, PRDX1, THBD
- Alport syndrome (NEF01v.16.1; 3 genes)**
COL4A3, COL4A4, COL4A5
- Alport syndrome, broad differential diagnosis (NEF23v21.1; 22 genes)**
ACTN4, C3, CD2AP, CFH, CFHR5, COL4A1, COL4A3, COL4A4, COL4A5, FAT1, FN1, INF2, ITGB4, LAMA5, LMX1B, MYH9, MYO1E, NPHS1, NPHS2, SLC7A7, TRPC6, WT1
- Chronic kidney disease of the young (CKD-Y) (includes PKD1 and PKD2) (NEF24v23.1; 260 genes)**
ACE, ACTG2, ACTN4, ADAMTS9, AGT, AGTR1, AGXT, AHI1, ALG1, ALMS1, AMN, ANKS6, ANLN, APOA1, APOA2, APOC2, APOE, APOL1, APRT, ARHGAP10, ARL13B, ARL6, ARMC9, ATXN10, AVIL, B2M, B9D1, B9D2, BBIIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCS1L, BMPR2, C1GALT1C1, C3, C8ORF37, CACNA1D, CACNA1H, CC2D2A, CD151, CD2AP, CD46, CDK20, CEP104, CEP164, CEP290, CEP41, CEP83, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CHD7, CLCN2, CLCN5, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CPLANE1, CRB2, CSPPT, CTNS, CUBN, CUL3, CYP11B1, CYP11B2, CYP17A1, DAAM2, DDC1, DDC2C, DGKE, DCL1, DNAJB11, DSTYK, E2F3, EMP2, EYA1, FAM149B1, FAN1, FAT1, FGA, FNAJ, FOXC2, FRAS1, FREM1, FREM2, GANAB, GAPVD1, GATA3, GATM, GLA, GLIS2, GRHRP, GRIP1, GSN, HNF1B, HOGA1, HPSE2, HSD11B2, HYL5, IFT27, IFT74, IFT81, IL1RAP, INF2, INPP5E, INVS, IQCB1, ITGA3, ITGA8, ITGB4, ITSN1, ITSN2, JAG1, KANK1, KANK2, KANK4, KATNIP, KCNJ5, KIAA0586, KIF3B, KIRREL1, KLHL3, LAMB2, LCAT, LMNA, LMX1B, LRIG2, LYZ, LZTFL1, MAFB, MAGI2, MAP7D3, MAPKBP1, MKKS, MKS1, MMACHC, MOCOS, MTR, MTRR, MTX2, MUC1, MYH11, MYH9, MYO1E, NEK8, NOS1AP, NOTCH2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NR3C1, NR3C2, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, NXF5, OCLRF, OFD1, OSGEP, PAX2, PBX1, PCM1, PDS1, PDS2, PIBF1, PKD1, PKD2, PKHD1, PLCE1, PMM2, POC1B, PODXL, PTPRO, REN, RMN1D, ROBO2, RPRGRI1L, RRM2B, SALL1, SARS2, SCARB2, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEC61A1, SGLP1, SIX1, SIX5, SLC22A12, SLC2A9, SLC3A1, SLC41A1, SLC4A1, SLC7A7, SLC7A9, SMARCAL1, SOX17, STX16, TBC1D8B, TBX18, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TMEM72, TNS2, TNXB, TOGARAM1, TP53RK, TPRKB, TRAF3IP1, TRAP1, TRIM32, TRIM8, TRPC6, TTC21B, TTC8, TTR, TULP3, UMOD, VIPAS39, VPS33B, WDR19, WDR35, WDR60, WDR73, WNK1, WNK4, WT1, XDH, XPNPEP3, YRDC, ZMPSTE24, ZNF423
In General the analysis will not detect MUC1 VNTR Cytosine-insertions.
- Copy number analysis: HNF1B NPHP1

- Congenital anomalies of the kidney and urinary tract (CAKUT) (NEF03v23.2; 119 genes)**

ACE, ACTA2, ACTG2, AGT, AGTR1, ANOS1, BMP4, BNC2, CBWD1, CENPF, CEP55, CHD1L, CHD7, CHRM3, CHRNA3, COQ7, CTU2, DACT1, DHCR7, DOCK4, DSTYK, EVX1, EYA1, FAM58A, FGF20, FGF8, FOXO1, FOXF1, FRAS1, FREM1, FREM2, GATA3, GDF6, GDNF, GFRA1, GLI3, GPC3, GREB1L, GREM1, GRIP1, HAAO, HNF1B, HOXA10, HOXA13, HOXD13, ...

In submitting this sample the clinician confirms that the patient has been informed about the chances of uncovering incidental findings that can result from this medical test.

⁸ Sequence and copy number analysis

[^] Repeat expansion analysis only

Renal disease (Continued)

Gene panels (incl. WES based CNV analysis per gene)

....HPSE2, HSPA6, ISL1, ITGA8, ITGB4, JAG1, KCTD1, KDM2B, KDM6A, KIF14, KMT2D, KYNU, LHX1, LIFR, LMOD1, LPP, LRIG2, LRP10, LRP4, MKKS, MYH11, MYLK, MYOCD, NAALADL2, NADSYN1, NCAPG2, NIPBL, NOTCH2, NPHP1, NPHP3, NPHP4, NPNT, NRIP1, PAX2, PAX8, PBX1, PLVAP, RBM8A, REN, RET, ROBO1, ROBO2, ROR2, SALL1, SALL4, SIX1, SIX2, SIX5, SKAP2, SLIT2, SLIT3, SOX11, SOX17, SRGAP1, STRA6, TBC1D1, TBX18, TBX6, TFAP2A, TMEM260, TNXB, TP63, TRAP1, TSHZ3, TXNL4A, UMOD, UPK3A, WBP11, WNT4, WNT9B, WT1, ZEB2, ZIC3, ZMYM2

Copy number analysis: EYA1 HNF1B

NPHP1 RET

Dents disease (type 1 and type 2) / Lowe syndrome / Cystinose (NEF22v16.2; 3 genes)

CLCN5, CTNS, OCRL

Diabetes insipidus, nephrogenic and neurogenic (NEF25v16.1; 3 genes)

AQP2, AVP, AVPR2

Electrolyte disorder (including Bartter syndrome, Gitelman syndrome and hypomagnesemia) (NEF09v23.1; 38 genes)

ATP1A1, BSND, CACNA1S, CASR, CLCN5, CLCNKA, CLCNKB, CLDN10, CLDN16, CLDN19, CNNM2, DGAT1, EGF, EPCAM, FXR2, GUCY2C, HNF1B, KCNJ1, KCNJ10, KCNJ16, MAGED2, MYO5B, NEUROG3, PCBD1, RRAGD, SARS2, SCN4A, SCNN1A, SCNN1B, SCNN1G, SLC12A1, SLC12A3, SLC26A1, SLC26A3, SLC41A1, SLC9A3, SPINT2, TRPM6

Copy number analysis: CLCNKB SLC12A3

Hereditary kidney disease full panel (NEF00v23.1; 527 genes including kidney tumor associated genes)

Pre-test genetic counselling required; please use NEF24 in case of kidney failure of unknown cause

In General the analysis will not detect MUC1 VNTR Cytosine-insertions.

ACE, ACTA2, ACTG2, ACTN4, ADAMTS13, ADAMTS9, ADCK3, ADCY10, AGK, AGT, AGTR1, AGXT, AHI1, ALDOB, ALG1, ALG5, ALG6, ALG8, ALG9, ALMS1, ALPL, AMN, ANKFY1, ANKS3, ANKS6, ANLN, ANOS1, AP2S1, APOA1, APOA2, APOC2, APOE, APOL1, APRT, AQP2, ARHGDI1, ARL13B, ARL3, ARL6, ARMC9, ARSA, ATP1A1, ATP6V0A4, ATP6V1B1, ATP7B, ATXN10, AVIL, AVP, AVPR2, B2M, B9D1, B9D2, BAP1, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCS1L, BMP4, BMPR2, BNC2, BSND, C1GALT1C1, C2CD3, C3, C8ORF37, CA2, CACNA1D, CACNA1H, CACNA1S, CASR, CBWD1, CBY1, CC2D2A, CCDC114, CCDC28B, CCN9, CD151, CD2AP, CD46, CDC73, CDK20, CDKN1C, CENPF, CEP104, CEP120, CEP164, CEP290, CEP41, CEP55, CEP83, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CHD1L, CHD7, CHRM3, CHRNA3, CLCN2, CLCN5, CLCNKA, CLCNKB, CLDN10, CLDN16, CLDN19, CNM2, COL4A1, COL4A3, COL4A4, COL4A5, COQ2, COQ4, COQ6, COO7, COO8B, COO9, COX10, CPLANE1, CPT2, CRB2, CRKL, CSPP1, CTNS, CTU2, CUBN, CUL3, CYP11B1, CYP11B2, CYP17A1, CYP24A1, CYP27B1, CYP2R1, CYP3A4, DAAM2, DACT1, DCDC2, DDX59, DGAT1, DGKE, DHCR7, DICER1, DLG1, DLG5, DMP1, DNAJB11, DOCK4, DST, DSTYK, DYNC2H1, DYNC2L1, DZIP1L, E2F3, EGF, EHHADH, ELP1, EMP2, ENPP1, EPCAM, ERCC6, ERCC8, EVC, EVC2, EVX1, EXOC8, EYA1, FAH, FAHD2A, FAM111A, FAM134B, FAM149B1, FAM20A, FAM20C, FAN1, FAT1, FBXL4, FGA, FGF20, FGF23, FGF8, FGFRL1, FH, FLCN, FN1, FOXC1, FOXC2, FOXF1, FOXI1, FRAS1, FREM1, FREM2, FXR2, G6PC, GALNT3, GALT, GANAB, GAPVD1, GATA3, GATM, GCM2, GDF6, GDNF, GFRA1, GLA, GLI3, GLIS2, GLIS3, GNA11, GNAS, GON7, GPC3, GPC5, GREB1L, GREM1, GRHRP, GRP1, GSN, GUCY2C, HAAO, HNF1A, HNF1B, HNF4A, HOGA1, HOXA10, HOXA13, HOXD13, HPR1, HPSE2, HRAS, HSD11B2, HSPA6, HYL51, ICK, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT57, IFT74, IFT80, IFT81, IL1RAP, INF2, INPP5E, INTU, INVS, IQCB1, ISL1, ITGA3, ITGA8, ITGB4, ITSN1, ITSN2, JAG1, KANK1, KANK2, KANK4, KATNIP, KCNJ1, KCNJ10, KCNJ16, KCNJ5, KCTD1, KCTD3, KDM2B, KDM6A, KIAA0586, KIAA0753, KIF14, KIF3B, KIF7, KIRREL1, KL, KLHL3, KMT2D, KRAS, KYNU, LAGE3, LAMA5, LAMB2, LCAT, LHX1, LIFR, LMNA, LMOD1, LMX1B, LPP, LRIG2, LRP10, LRP2, LRP4, LRP5, LYZ, LZTFL1, MAFB, MAGED2, MAGI2, MAP7D3, MAPKBP1, MET, MKKS, MKS1, MMACHC, MOCOS, MTR, MTRR, MTX2, MUC1, MYH11, MYH9, MYLK, MYO1E, MYO5B, MYOCD, NAALADL2, NADSYN1, NCAPG2, NEK1, NEK8, NEU1, NEUROG3, NGF, NIPBL, NOS1AP, NOTCH2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NPNT, NR3C1, NR3C2, NRAS, NRIP1, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, NXF5, OCRL, OFD1, OSGEP, OXGR1, PAX2, PAX8, PBX1, PCBD1, PCM1, PDE6D, PDS1, PDS2, PHEX, PIBF1, PKD1, PKD2, PKHD1, PLVAP, PMM2, POC1B, PODXL, PRDM12, PRDX1, PRKCSH, PSAP, PTEN, PTH1R, PTPRO, PYGM, RBM8A, REN, RERE, RET, RICTOR, RIMD1, ROBO1, ROBO2, ROR2, RRGRI1, RRGRI1P, RRAGD, RRM2B, SALL1, SALL4, SARS2, SCARB2, SCLT1, SCN11A, SCN4A, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SDHB, SEC61A1, SEC61B, SEC63, SGPL1, SIX1, SIX2, SIX5, SKAP2, SLC12A1, SLC12A3, SLC16A12, SLC19A2, SLC22A12, SLC26A1, SLC26A3, SLC2A2, SLC2A9, SLC34A1, SLC34A3, SLC36A2, SLC37A4, SLC3A1, SLC41A1, SLC4A1, SLC4A4, SLC5A2, SLC6A19, SLC6A20, SLC7A7, SLC7A9, SLC9A3, SLC9A3R1, SLIT2, SLIT3, SMARCAL1, SOX11, SOX17, SPINT2, SPTLC1, SPTLC2, SRGAP1, STRA6, STRADA, STX16, SUFU, SYNPO, TBC1D1, TBC1D8B, TBCE, TBX18, TBX6, TCTEX1D2, TCTN1, TCTN2, TCTN3, TFAP2A, THBD, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM260, TMEM67, TMEM72, TNS2, TNXB, TOGARAM1, TP53RK, TP63, TPRKB, TRAF3IP1, TRAP1, TRIM32, TRIM8, TRPC6, TRPM6, TRPM7, TSC1, TSC2, TSHZ3, TTC21B, TTC8, TTR, TUBB4B, TULP3, TXNDC15, TXNL4A, UMOD, UPK3A, UQC2, VDR, VHL, VIPAS39, VPS33B, WBP11, WDR39, WDR35, WDR60, WDR72, WDR73, WNK1, WNK4, WNT4, WNT9B, WT1, XDH, XPNPEP3, XPO5, YRDC, ZEB2, ZIC3, ZMPSTE24, ZMYM2, ZNF365, ZNF423

Hypertension / Pseudohypoaldosteronism (NEF15v21.1; 21 genes)

BMPR2, CACNA1D, CACNA1H, CLCN2, CUL3, CYP11B1, CYP11B2, CYP17A1, HSD11B2, KCNJ5, KLHL3, MTX2, NR3C1, NR3C2, SARS2, SCNN1A, SCNN1B, SCNN1G, STX16, WNK1, WNK4

Hyperuricemia / Uricosuria (NEF08v21.1; 14 genes)

ALDOB, ATP7B, CTNS, G6PC, GALT, HPR1, MOCOS, PYGM, REN, SARS2, SLC22A12, SLC2A9, SLC37A4, UMOD

Nephrocalcinosis / Nephrolithiasis (NEF10v23.1; 66 genes)

ADCY10, AGK, AGXT, ALDOB, AP2S1, APRT, ATP6V0A4, ATP6V1B1, ATP7B, BSND, CA2, CASR, CLCN5, CLCNKB, CLDN10, CLDN16, CLDN19, CTNS, CYP24A1, DMP1, ENPP1, FAM20A, FGF23, FOXI1, G6PC, GALT, GNA11, GRHRP, HNF4A, HOGA1, HPR1, KCNJ1, KCNJ16, MAGED2, MOCOS, OCRL, OXGR1, PHEX, PTH1R, SCNN1A, SCNN1B, SCNN1G, SLC12A1, SLC22A12, SLC26A1, SLC2A9, SLC34A1, SLC34A3, SLC36A2, SLC37A4, SLC3A1, SLC4A1, SLC6A19, SLC6A20, SLC7A9, SLC9A3R1, STRADA, STX16, TRPM6, VDR, VIPAS39, VPS33B, WDR72, WNK4, XDH, ZNF365

Copy number analysis: SLC3A1 SLC7A9

Nephrotic syndrome (NPHS) / Focal segmental glomerulosclerosis (FSGS) (NEF11v23.1; 106 genes)

ACTN4, ADCK3, ALG1, ALMS1, AMN, ANKFY1, ANLN, APOA1, APOE, APOL1, ARHGDI1, AVIL, B2M, CD151, CD2AP, CDK20, CFH, CLCN5, COL4A3, COL4A4, COL4A5, COQ2, COQ4, COQ6, COQ7, COQ8B, COQ9, CRB2, CUBN, DAAM2, DGKE, DLG1, E2F3, EMP2, ERCC6, ERCC8, FAT1, FGA, FN1, FOXC2, GAPVD1, GLA, GON7, GPC5, GSN, HNF1B, IL1RAP, INF2, ITGA3, ITGB4, ITSN1, ITSN2, KANK1, KANK2, KANK4, KIRREL1, LAGE3, LAMA5, LAMB2, LCAT, LMNA, LMX1B, LYZ, MAFB, MAGI2, MTR, MYH9, MYO1E, NOS1AP, NPHP4, NPHS1, NPHS2, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, NXF5, OCRL, OSGEP, PAX2, PDS1, PDS2, PLCE1, PMM2, PODXL, PTPRO, SCARB2, SEC61A1, SGPL1, SLC7A7, SMARCAL1, SYNPO, TBC1D8B, TNS2, TP53RK, TPRKB, TRIM8, TRPC6, TTC21B, WDR73, WT1, XPO5, YRDC, ZMPSTE24

Renal cysts and/or ciliopathies, incl. Bardet-Biedl syndrome, Nephronophthisis and Joubert syndrome (NEF17v23.2; 165 genes)

ADAMTS9, AGXT, AHI1, ALG5, ALG6, ALG8, ALG9, ALMS1, ANKS3, ANKS6, ARL13B, ARL3, ARL6, ARMC9, ATXN10, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C2CD3, C8ORF37, CBY1, CC2D2A, CCDC114, CCDC28B, CDC73, CDKN1C, CENPF, CEP104, CEP120, CEP164, CEP290, CEP41, CEP55, CEP83, CLDN10, COL4A1, COL4A3, COL4A4, COL4A5, CPLANE1, CPT2, CRB2, CSPP1, DCDC2, DDX59, DHCR7, DICER1, DNAJB11, DYNC2H1, DYNC2L1, DZIP1L, EVC, EVC2, EXOC8, FAM149B1, FAN1, GANAB, GLIS2, GLIS3, GPC3, HNF1B, HYL51, ICK, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT57, IFT74, IFT80, IFT81, INPP5E, INTU, INVS, IQCB1, JAG1, KATNIP, KIAA0586, KIAA0753, KIF14, KIF3B, KIF7, LRP5, LZTFL1, MAP7D3, MAPKBP1, MKKS, MKS1, MUC1, NCAPG2, NEK1, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, OFD1, PBX1, PCM1, PDE6D, PIBF1, PKD1, PKD2, PKHD1, PMM2, POC1B, PRKCSH, REN, RERE, RIMD1, RRGRI1, RRGRI1P, SCLT1, SDCCAG8, SEC61A1, SEC61B, SEC63, SLC4A1, SLC4A1, TBX18, TCTEX1D2, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TMEM72, TOGARAM1, TRAF3IP1, TRIM32, TSC1, TSC2, TTC21B, TTC8, TXNDC15, UMOD, VHL, WDR39, WDR72, WDR73, WDR35, WDR60, XPNPEP3, ZIC3, ZNF423, ATP6V0A4, ATP6V1B1, DLG5, PAX2, SALL1, SUFU, TMEM218, TULP3

In General the analysis will not detect MUC1 VNTR Cytosine-insertions.

Copy number analysis: HNF1B NPHP1

Renal cysts in adulthood / autosomal dominant tubulointerstitial kidney disease (ADTKD) (NEF26v23.1; 38 genes)

ALG5, ALG6, ALG8, ALG9, ATP6V0A4, ATP6V1B1, CDC73, COL4A1, COL4A3, COL4A4, COL4A5, DNAJB11, GANAB, HNF1B, IFT140, JAG1, LRP5, MAPKBP1, MUC1, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, OFD1, PBX1, PCM1, PDE6D, PIBF1, PKD1, PKD2, PKHD1, PMM2, POC1B, PRKCSH, REN, SALL1, SEC61A1, SEC61B, SEC63, SLC4A1, TSC1, TSC2, UMOD, VHL

In General the analysis will not detect MUC1 VNTR Cytosine-insertions.

Renal Fanconi Syndrome (NEF16v23.1; 34 genes)

ALDOB, AMN, ARSA, ATP7B, BCS1L, CLCN5, COQ7, COQ9, COX10, CTNS, CUBN, EHHADH, FAH, FAHD2A, G6PC, GALT, GATM, GLA, HNF4A, LRP2, OCRL, PSAP, RIMD1, SLC16A12, SLC19A2, SLC26A1, SLC2A2, SLC34A1, SLC37A4, SLC5A2, SLC6A19, SLC6A20, VIPAS39, VPS33B

Renal phosphate-handling (NEF18v21.1; 24 genes)

ALPL, CLCN5, CYP27B1, CYP2R1, CYP3A4, DMP1, ENPP1, FAH, FAM20C, FGF23, GFR1, GALNT3, GATM, KCNJ1, SLC12A1, SLC12A3, SLC37A4, PHEX, SLC34A1, SLC34A3, SLC9A3R1, VDR

Renal Tubular Acidosis (NEF19v21.1; 22 genes)

ATP6V0A4, ATP6V1B1, BSND, CA2, CLCNKB, COQ9, EHHADH, FBXL4, FN1, FOXI1, G6PC, GATM, KCNJ1, SLC12A1, SLC12A3, SLC37A4, SLC4A1, SLC4A4, UQC2, VIPAS39, VPS33B, WDR72

In submitting this sample the clinician confirms that the patient has been informed about the chances of uncovering incidental findings that can result from this medical test.

® Sequence and copy number analysis

^ Repeat expansion analysis only

Renal disease (Continued)

Gene panels (incl. WES based CNV analysis per gene)

- Renal Tubular Dysgenesis (NEF20v16.1; 5 genes)**
ACE, AGT, AGTR1, REN, UMOD
- Chronic kidney disease-kids, CKD-kids (including PKD1 and PKD2) (NEF27 v23.1; 360 genes)**
ACE, ACTG2, ACTN4, ADAMTS9, AGT, AGTR1, AGXT, AHI1, ALG1, ALG5, ALG6, ALG8, ALG9, ALMS1, AMN, ANKFY1, ANKS3, ANKS6, ANLN, ANOS1, APOA1, APOA2, APOC2, APOE, APOL1, APRT, ARHGDI1, ARL13B, ARL3, ARL6, ARMC9, ATP6V0A4, ATP6V1B1, ATXN10, AVIL, B2M, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCS1L, BMPR2, BNC2, C1GALT1C1, C2CD3, C3, CACNA1D, CACNA1H, CBY1, CC2D2A, CCDC28B, CCNQ, CD151, CD2AP, CD46, CDC73, CDK20, CDKN1C, CENPF, CEP104, CEP120, CEP164, CEP290, CEP41, CEP55, CEP83, CFAP418, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CHD1L, CHD7, CHRNA3, CILK1, CLCN2, CLCN5, CLDN10, COL4A1, COL4A3, COL4A4, COL4A5, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, CPLANE1, CPT2, CRB2, CSPP1, CTNS, CUBN, CUL3, CYP11B1, CYP11B2, CYP17A1, DAAM2, DACT1, DCDC2, DDX59, DGKE, DHCR7, DICER1, DLC1, DLG5, DNAJB11, DSTYK, DYNC2H1, DYNC211, DYNC212, DYNC2L11, DYNL2B, DZIP1L, E2F3, EMP2, ERCC6, ERCC8, EVC, EVC2, EXOC8, EYA1, FAM149B1, FAN1, FAT1, FGA, FGF20, FN1, FOXC2, FRAS1, FREM1, FREM2, GANAB, GAPVD1, GATA3, GATM, GDF6, GFRA1, GLA, GLIS2, GLIS3, GON7, GPC3, GPC5, GREB1L, GRHRP, GRIP1, GSN, HNF1B, HOGA1, HOXA13, HPSE2, HSD11B2, HYL5L1, IFT122, IFT140, IFT172, IFT43, IFT52, IFT57, IFT74, IFT80, IFT81, IL1RAP, INF2, INPP5E, INTU, INVS, IQCB1, ITGA3, ITGA8, ITGB4, ITSN1, ITSN2, JAG1, KANK1, KANK2, KANK4, KATNIP, KCNJ5, KCTD1, KIAA0586, KIAA0753, KIF14, KIF3B, KIF7, KIRREL1, KLHL3, LAGE3, LAMA5, LAMB2, LCAT, LIFR, LMNA, LMOD1, LMX1B, LRIG2, LRP5, LYZ, LZTFL1, MAFB, MAGI2, MAP7D3, MAPKBP1, MKKS, MKS1, MMACHC, MOCOS, MTR, MTRR, MTX2, MUC1, MYH11, MYH9, MYLK, MYO1E, MYOCD, NADSYN1, NACAPG2, NEK1, NEK8, NOS1AP, NOTCH2, NPH1, NPH3, NPH4, NPHS1, NPHS2, NPNT, NR3C1, NR3C2, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, NXF5, OCRL, ODAD1, OFD1, OSGEP, PAX2, PBX1, PCM1, PDE6D, PDSS1, PDSS2, PIBF1, PKD1, PKD2, PKHD1, PLCE1, PMM2, POC1B, PODXL, PRKCSH, PTPRO, REN, RERE, RMND1, ROBO1, ROBO2, RPGRIP1, RPGRIP1L, RRM2B, SALL1, SARS2, SCARB2, SCLT1, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEC61A1, SEC61B, SEC63, SGPL1, SIX1, SIX5, SLC22A12, SLC2A9, SLC3A1, SLC41A1, SLC4A1, SLC7A7, SLC7A9, SLIT2, SMARCAL1, SOX17, STX16, SUFU, SYNPO, TBC1D1, TBC1D8B, TBX18, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TMEM72, TNS2, TNXB, TOGARAM1, TP53RK, TP63, TPRKB, TRAF3IP1, TRAP1, TRIM32, TRIM8, TRPC6, TSC1, TSC2, TTC21B, TTC8, TTR, TULP3, TXNDC15, UMOD, UPK3A, VHL, VIPAS39, VPS33B, WDPCP, WDR19, WDR35, WDR73, WNK1, WNK4, WNT9B, WT1, XDH, XPNPEP3, XPO5, YRDC, ZIC3, ZMPSTE24, ZNF423, ZNG1A
In General the analysis will not detect MUC1 VNTR Cytosine-insertions.

- Hereditary angioedema (HAE01v21.1; 7 genes)**
ANGPT1, F12, HS3ST6, KNG1, MYOF, PLG, SERPING1
- Familial partial lipodystrophy (FPLD) and congenital generalized lipodystrophy (CGL) (LIP01v22.1; 11 genes)**
AGPAT2, AKT2, BSCL2, CAV1, CIDEC, LIPE, LMNA, PLIN1, PPARG, PTRF, ZMPSTE24
- Idiopathic pulmonary fibrosis (IPF01v21.1; 34 genes)**
ABCA3, ACD, AP3B1, COPA, CSF2RA, CSF2RB, DKC1, DTNBP1, FAM111B, FARSA, FARSB, HPS1, HPS4, ITGA3, MARS, NHP2, NKX2-1, NOP10, OAS1, PARN, POT1, RTEL1, SFTPA1, SFTPA2, SFTPB, SFTPC, TERC, TERT, TINF2, TMEM173, USB1, WRAP53, ZCCHC8, FOXF1
- Neonatal erythroderma (ERY00v17.1; 60 genes)**
ABCA12, ABHD5, ADAM17, ALDH3A2, ALOX12B, ALOXE3, ASS1, ATP7A, BCKDHA, BCKDHB, BTD, BTK, C5, C8A, C8B, C8G, CARD14, CLDN1, CPS1, CYP4F22, CERS3, CDSN, DCLRE1C, DSG1, DBT, DLD, EBP, ELOVL4, ERCC2, ERCC3, GBA, GJB2, GJB6, GTF2H5, HLCS, IL36RN, KIT, KRT1, KRT10, KRT2, LIPN, LOR, MPLKIP, MBTPS2, MUT, NIPAL4, NSDHL, PCCA, POMP, PNPLA1, PCCB, RAG1, RAG2, STST, SLC25A13, SLC30A2, SLC39A4, SPINK5, TBX1, TGM1
- Nonsyndromal disorders of sex development* (DSD) (DSD00v21.1; 38 genes)**
Includes copy number analysis of SRY, SOX9, NR0B1, SOX3
AKR1C2, AKR1C4, AMH, AMHR2, AR, CBX2, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, DHH, DHX37, DMRT1, DMRT2, ESR2, HSD17B3, HSD3B2, LHB, LHCGR, MAMLD1, MAP3K1, NR0B1, NR2F2, NR3C1, NR5A1, POR, PSMC3IP, RSP01, SOX3, SOX9, SRD5A2, SRY, STAR, TSPYL1, WNT4, WT1, ZFPM2
Repeat expansion analysis*: AR
- Syndromal disorders of sex development* (DSD) (DSD01v21.2; 134 genes)**
Includes copy number analysis of SRY, SOX9, NR0B1, SOX3
AIRE, AKR1C2, AKR1C4, AMH, AMHR2, ANOS1, AR, ARMC5, ATRX, B9D1, BMP15, CBX2, CCNQ, CDKN1C, CEP41, CHD7, CILK1, CLPP, CUL4B, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR7, DHH, DHX37, DMRT1, DMRT2, DUSP6, DYNC2H1, EIF2B5, ERAL1, ESR1, ESR2, FEZF1, FGF17, FGF8, FGFR1, FGFR2, FLRT3, FOXL2, FRAS1, FREM2, FSHB, FSHR, FZD2, GATA4, GDF9, GK, GLI2, GNRH1, GNRHR, GRIP1, HESX1, HFM1, HHAT, HOXA13, HS6ST1, HSD17B3, HSD17B4, HSD3B2, IL17RD, INPP5E, IRF6, KISS1, KISS1R, LARS2, LEP, LEPR, LHB, LHCGR, LHX3, MAMLD1, MAP3K1, MCM5, MCM8, MCM9, MKKS, MKRN3, MYRF, NEK1, NNT, NOBOX, NR0B1, NR2F2, NR3C1, NR5A1, NSMF, PBX1, PCSK1, PLXNA1, PNPLA6, POLE, POR, PPP1R12A, PROK2, PROKR2, PROP1, PSMC3IP, RPKA4, ROR2, RPL10, RSP01, SAMD9, SEMA3A, SEMA3E, SGPL1, SOHLH1, SOX10, SOX2, SOX3, SOX8, SOX9, SPRY4, SRAP, SRD5A2, SRY, STAG3, STAR, SYCE1, TAC3, TACR3, TBX3, TOE1, TSPYL1, TWIST2, TWNK, WDR11, WDR60, WNT4, WT1, ZFPM2
Repeat expansion analysis*: AR

Renal disease

Single gene | Sequence analysis

- Gitelman syndrome[§] SLC12A3[§]
- Glomerulopathy with fibronectin deposition (GFND2) FN1
- APOL1-mediated kidney disease, risk factor APOL1
Applies only to familial diagnosis for G1/G2 risk allele
- Hypertension and brachydactyly syndrome/Bilginturan syndrome PDE3A
- Hypoparathyroidy, deafness and renal dysplastic syndrome GATA3
- Interstitial lung fibrosis and congenital nephrotic syndrome ITGA3

Other diseases

Gene panels (incl. WES based CNV analysis per gene)

- Amyloidosis (AMY01v19.1; 12 genes)**
APOA1, APOA2, APOC2, APOC3, B2M, CST3, FGA, GSN, IL31RA, LYZ, OSMR, TTR
- Congenital diarrhoea (DIA00v23.1; 73 genes)**
ADA, ADAM17, AIRE, ALPI, ANGPTL3, ANKZF1, APOB, CD3D, CD3E, CFTR, CLMP, CYP27A1, DCLRE1C, DGAT1, EPCAM, FLNA, FDXP3, GUCY2C, IL10, IL10RA, IL10RB, IL12RB1, IL21, IL2RA, IL2RG, IL7R, JAK3, LCT, LIPA, MPI, MTPP, MVK, MYO5B, NCF2, NCF4, NEUROG3, NHEJ1, NPC1L1, PCSK1, PCSK9, PERCC1, PLVAP, PNPLP, PNP, PRSS1, PRSS2, PTPRC, RAG1, RAG2, SAR1B, SBDS, SI, SKIV2L, SLC10A2, SLC26A3, SLC2A2, SLC39A4, SLC5A1, SLC7A7, SLC9A3, SPINK1, SPINT2, STAT1, STAT5B, STX3, TCN2, TMPRSS15, TTC37, TTC7A, UBR1, WNT2B, XIAP, ZAP70

Other diseases

Single gene | Sequence analysis

- Azoö/oligozoöspermie (AZF) (only deletion/duplication test) [AZF]
- Amyloidosis I en VII; transthyretin amyloidosis TTR
- Diarrhea 2, with microvillus atrophy (DIAR2)[§] MYO5B[§]
- Fragile X tremor/ataxia syndrome (FXTAS)[^] FMR1[^]
- Premature ovarian failure, (POF1)[^] FMR1[^]
- Surfactant metabolism dysfunction type 3 (SMDP3) ABCA3
- Uniparental disomy, chromosome:..... [MARK]
- X-chromosome inactivation AR
- 15q11-q13 duplication syndrome (methylation sensitive deletion/duplication test) [15q11-q13]

In submitting this sample the clinician confirms that the patient has been informed about the chances of uncovering incidental findings that can result from this medical test.

[§] Sequence and copy number analysis

[^] Repeat expansion analysis only

Genome Diagnostics Section

Department of Genetics
University Medical Center (UMC) Utrecht
Heidelberglaan 100
3584 CX Utrecht

**PATIENT COPY****Use of patient material**

You have provided a sample (e.g. blood, bone marrow, urine, skin biopsy, buccal tissue, amniotic fluid) for DNA testing. Your DNA will be investigated for a possible cause of your condition. It is the responsibility of your physician to inform you on the testing procedure(s), benefits and limitations of the test(s) and possible consequences of the test results. During testing we typically only use part of the DNA we extracted from your sample. The rest of the DNA, the leftover, is stored for at least thirty years according to the guidelines of the professional association of clinical genetic laboratory specialists (VKGL) and is available for future DNA testing on your behalf.

- Providing up-to date genetic diagnostic testing requires ongoing improvement, development and implementation of (new) analysis methods and techniques. The usage of anonymised (de-identified) leftover patient DNA is vital for these improvements. When using your leftover DNA, we comply to the rules of conduct set by the Dutch Federation of Medical Scientific Societies (FMWV): www.federa.org.

- The UMC Utrecht is an academic institution. Its task is to innovate and improve healthcare and to conduct medical scientific research. Your rights and privacy are governed by UMC Utrecht regulations. For detailed information about privacy and the protection of personal data, we refer to the website of the UMC Utrecht: Practical > Rights and regulations > Use of residual material. Permission to use residual material can also be changed here. (see <https://www.umcutrecht.nl/nl/Ziekenhuis/In-het-ziekenhuis/Regels-en-rechten/Gebruik-lichaamsmateriaal-medische-gegevens/Bezwaarformulier>)

Complaints

At the UMC Utrecht we strive to provide the best possible care. If you are unhappy it is often worthwhile discussing your concerns early on with your physician. However, if you do not feel comfortable raising your concerns directly or your problem was not resolved you can contact the UMC Utrecht complaints mediation service. The complaints mediators mediate in patient complaints about the hospital and are also able to help you submit your complaint. The complaints mediators can be contacted via the UMC Utrecht website: www.umcutrecht.nl.

Please contact your referring physician to discuss any questions you may have.



The genome diagnostics section has been certified with
NEN-EN-ISO 15189:2012 by the Accreditation Council.
The scope of accreditation number M001 can be seen on
www.rva.nl.