SEND TO
Genome Diagnostics Section
University Medical Center Utrecht
Centrale Balie CDL
Huispost G.03.3.30
Heidelberglaan 100
3584 CX Utrecht
The Netherlands

UMC Utrecht

Laboratory Opening Hours :8:30-17:00 Mon-Fri
Tel +31 (0)88 – 75 54090
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 excisting 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

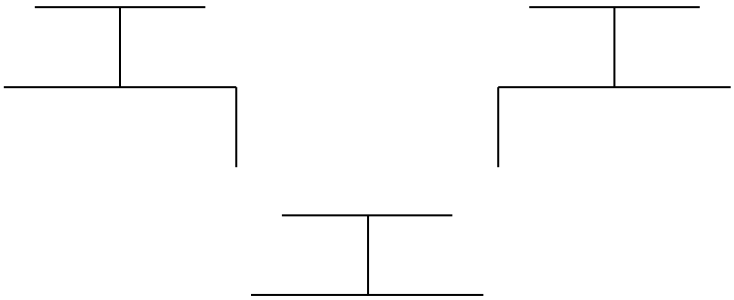
Datum:

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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Tests available

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.

TESTEN RvA M 001

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, MPIG6B, 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, TBXA2R, 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 syndroom** (ROW01v22.1; 4 genes)

Including deletion/duplicationtest ENG & ACVRL1

ENG, ACVRL1, GDF2, SMAD4

Blood disorders and vascular disease
Single gene | Sequence analysis

☐ G6PD deficientie	G6PD
☐ Haemophilia A, (HEMA)[δ]	F8[δ]
☐ Von Willebrand Factor	VWF

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

☐ **Cardiomyopathy** (CAR01v24.1; 46 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, CACNA1C, CRYAB, CSRP3, DES, DMD, DSC2, DSG2, DSP, FHL1, FHOD3, FLNC, GLA, HCN4, JPH2, JUP, KLHL24, LAMP2, LMNA, MIB1, MYBPC3, MYH7, MYL2, MYL3, NEXN, PKP2, PLN, PRDM16, PRKAG2, RBM20, RYR2, SCN5A, TAZ, TCAP, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, 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 polymorfic 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, GGCX, 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.

[δ] 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, TGFBR1, TGFBR2, THSD4

☐ **Idiopatic 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, TNNI3, 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** (OWS01v24.1; 91 genes + 1 region (Chr22q11.2))
Includes copy number analyses for EYA1
BMP4, BMP5, BUB3, CDC45, CDC6, CDH11, CDT1, CHD7, DACT1, DCHS1, DDX59, DHODH, DHX37, DONSON, DRG1, EDNRA, EFNB1, EFTUD2, EIF4A3, EYA1, FANCB, FANCF, FANCL, FAT4, FBXL7, FBXO11, FGF10, FGF3, FGFR1, FOXI3, FRAS1, FREM2, FRK, GDF6, GMNN, GNAI3, GSC, HMX1, HOXA2, HSPA9, HUWE1, ITPR1, KCTD1, KDM6A, KMT2D, LAMA5, MARS1, MCM5, MED12, MED16, NF1, NID2, NRP1, OFD1, ORC1, ORC4, ORC6, OTX2, PAX1, PIK3CA, PLCB4, PLCD3, POLR1A, POLR1B, POLR1C, POLR1D, POMT1, PORCN, RBM10, RECQL, RECQL4, ROBO1, RPS26, RPS28, SALL1, SALL4, SF3B2, SF3B4, SIX1, SIX5, STAG2, TBX1, TCOF1, TFAP2A, TPRN, TSHZ1, TSR2, TXNL4A, WBP11, ZIC3, ZYG11B

☐ **(Non)syndromaal cleft lip and/or palate incl. Robin sequence** (OWS02v24.1; 203 genes + 1 region (Chr22q11.2))
Pretest counceling by clinical geneticist is required
ACTB, ACTG1, ALX1, ALX3, AMER1, AMMECR1, AMOTL1, ANKRD11, ARHGAP29, ARHGAP31, ASXL1, B3GALT6, B3GALTL, B4GALT7, B9D2, BCOR, BMP2, BMPER, C2CD3, C5orf42, CAMTA1, CC2D2A, CCDC32, CDC45, CDH1, CDKN1C, CHD7, CHRNG, CHST14, COL11A1, COL11A2, COL2A1, COL9A1, COLEC10, COLEC11, CTCF, CTNND1, DDX3X, DDX59, DHCR7, DHODH, DLL4, DOCK6, DVL1, DVL3, DYNC2H1, DYNC2LI1, 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, HYLS1, 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, PIGO, PIGV, PLCB4, POLR1A, POLR1C, POLR1D, POMT1, PORCN, PQBP1, PROKR2, PRRX1, PTCH1, PTCH2, PVRL1, RBM10, RIPK4, ROR2, RPGRIP1L, RPL11, RPL26, RPL5, RPS19, RPS26, RPS28, RUNX2, SALL4, SATB2, SCARF2, SEC23A, SEMA3E, SEPTIN9, SF3B4, SHH, SIX1, SIX3, SIX5, SKI, SLC10A7, SLC26A2, SMAD3, SMAD4, SMC1A, SMC3, SMCHD1, SMS, SNRPB, SON, SOX9, SPECC1L, STAC3, STAMBP, TAPT1, TBC1D32, TBX1, TBX15, TBX2, TBX22, TBX4, TCOF1, TCTN3, TFAP2A, TGDS, TGFB3, TGFBR1, TGFBR2, TGIF1, TMCO1, TMEM216, TMEM8C, TP63, TRIM37, TRRAP, 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 syndroom	ABCC9
☐ Cleidocraniale dysplasie (CCD)[δ]	RUNX2[δ]
☐ 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, ARHGEF9, 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, CHRNB2, CIC, CLCN4, CLN3, CLN5, CLN6, CLN8, CLTC, CNKSR2, CNNM2, CNPY3, CNTNAP2, COQ2, COQ4, CPLX1, CPT2, CSNK2B, CSTB, CTSD, CUL4B, CUX2, CYFIP2, D2HGDH, DCX, DEAF1, DENND5A, DEPDC5, DHDDS, DHPS, DIAPH1, DMXL2, DNAJC5, DNM1, DNM1L, 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, HCFC1, HCN1, HECW2, HNRNPR, HNRNPU, HSD17B10, HUWE1, INTS8, IQSEC2, IRF2BPL, KANSL1, KARS1, KAT8, KCNA1, KCNA2, KCNB1, KCNC1, KCNC2, KCND3, KCNH1, KCNJ10, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD3, KCTD7, KDM5C, NEXMIF, KIF1A, KMT2A, KPNA7, LGI1, LIAS, MAST3, MBD5, MBOAT7, MDH2, MECP2, MED12, MEF2C, MFSD8, MOCS1, MOCS2, MPDU1, MTHFR, MTOR, NACC1, NAPB, NBEA, NHLRC1, NPRL2, NPRL3, NRXN1, NSDHL, NTRK2, NUS1, OFD1, OPHN1, OTUD6B, PACS2, PAK1, PAK3, PARS2, PCDH19, PGAP1, PHACTR1, PHF6, PHGDH, PIGA, PIGB, PIGC, PIGG, PIGH, PIGN, PIGO, PIGP, PIGQ, 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, STXBP1, 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** (EPI04v24.1; 24 genes)
ATP1A2, CACNA1A, CHRNA2, CHRNA4, CHRNB2, 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, KCNMA1, 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.

[δ] Sequence and copy number analysis

^ Repeat expansion analysis only

Epilepsy
Individual genes | Sequence analysis

☐ Dravet syndrome (SMEI/SMEB)[δ]	SCN1A[δ]
☐ 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)
NB: Requests for this panel are reserved exclusively for clinical geneticists and via mainstreaming procedure
BRCA1 copynumber analysis included
BRCA1, BRCA2, ATM, BARD1, CHEK2, PALB2, RAD51C, RAD51D, PTEN, BRIP1

☐ **Ovarian cancer** (ONC01v22.1; 6 genes)
NB: Requests for this panel are reserved exclusively for clinical geneticists and via mainstreaming procedure
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)
NB: Requests for this panel are reserved exclusively for clinical geneticists
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)
NB: Requests for this panel are reserved exclusively for clinical geneticists
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)
NB: Requests for this panel are reserved exclusively for clinical geneticists
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)
NB: Requests for this panel are reserved exclusively for clinical geneticists and via mainstreaming procedure
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, CEBPA, CEP57, CREBBP, CTC1, CTLA4, 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, TINF2, 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)[δ]	MLH1[δ]
☐ Lynch syndrome (HNPCC1)[δ]	MSH2[δ]
☐ Lynch syndrome (HNPCC5)[δ]	MSH6[δ]
☐ Multiple Endocrine Neoplasia type 1 (MEN1)[δ]	MEN1[δ]
☐ Multiple Endocrine Neoplasia type 2 (MEN2) *(only relevant exons)*	RET
☐ Medullary thyroic cancer, sporadic (SMTC) *(on tumor tissue derived DNA only)*	RET
☐ Von Hippel-Lindau, disease (VHL)[δ]	VHL[δ]

Intellectual disability: syndromal/non-syndromal
Gene panel | Exome (incl. WES based CNV analysis per gene)

<u>This gene panel, and the exome-wide analysis, can only be requested by clinical geneticists.</u> 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[δ]	VPS13B[δ]
☐ Fragile-X syndrome, FRAXA ^	FMR1^
☐ Lesch-Nyhan syndrome, (LNS)	HPRT1
☐ Rett syndrome, RTT[δ]	MECP2[δ]
☐ Rett syndrome, atypical[δ]	CDKL5[δ]
☐ Rett syndrome, congenital variant[δ]	FOXG1[δ]
☐ 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.

[δ] 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 genen)
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, SLCO1B1, SLCO1B3, 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
☐ Familiar 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)[δ], *including deletion test exon 18*	GAA[δ]
☐ Pyruvaat Kinase deficiency (PK)	PKLR
☐ Tyrosinemia, type I	FAH
☐ Wilson disease (WD)	ATP7B

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•** (NEU01v24.2; 28 genes)
Includes repeat expansion analysis SCA2 (ATXN2) & C9ORF72
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

☐ **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

☐ **Moyamoya** (NEU06v24.1; 21 genes and region Xq28)
ACTA2, ANO1, BRCC3, CBL, CHD4, CNOT3, DIAPH1, GUCY1A3, JAG1, MTCP1, MTFMT, MYH11, NF1, NOS3, PTPN11, RASA1, RNF213, SAMHD1, SETD5, SHOC2, YY1AP1

Neurological disorders
Single gene | Sequence / repeat expansion 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, CHRNB1, CHRND, CHRNE, CHRNG, CLCN1, CLN3, CNTN1, CNTNAP1, COA7, COL12A1, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, COX6A1, CPT2, CRYAB, CTDP1, CWF19L1, CYP2U1, CYP7B1, DAG1, DCAF8, DCTN1, DDHD1, DDHD2, DES, DGAT2, DHTKD1, DMD, DNAJB2, DNAJB6, DNM2, DNMT1, DOK7, DPAGT1, DPM1, DPM2, DPM3, DST, DYNC1H1, DYSF, EBF3,ECEL1, EEF2, EGR2, ELOVL4, ELOVL5, ELP1, EMD, ENO3, ENTPD1, ERBB3, ERLIN1, ERLIN2, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, FA2H, FAM111B, RETREG1, FARS2, FASTKD2, FBLN5, FBXO38, FGD4, FGF14, FHL1, FIG4, FKRP, 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,

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.

[δ] Sequence and copy number analysis

^ Repeat expansion analysis only

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

KCNE5, KCNE2, KCNE3, KCNJ18, KCNJ2, KCNJ5, WASHC5, KIDINS220, KIF1A, KIF1B, KIF1C, KIF21A, KIF5A, KLC2, KLHL40, KLHL41, KLHL9, KY, L1CAM, LAMA2, LAMB2, LAMP2, LARGE1, 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, MYBPC3, MYF6, MYH2, MYH3, MYH7, MYH8, MYL1, MYL2, MYMK, MYO18B, MYO9A, MYOT, MYPN, NAGLU, NDRG1, NEB, NEFH, NEFL, NGF, NIPA1, NOP56, NT5C2, NTRK1, OBSCN, OPA1, ORAI1, PABPN1, PAX7, PDK3, PDYN, PEX7, PFKM, PGAM2, PGK1, PGM1,PHKA1, PHOX2A, PHYH, PIP5K1C, PLEC, PLEKHG5, PLP1, PMP2, PMP22, PNKP, PNPLA2, PNPLA6, PNPLA8, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, PRDM12, PREPL, PRKAG2, PRKCG, PRPS1, PRX, HACD1, CAVIN1, PTRH2, PUS1, PYGM, PYROXD1, RAB7A, RAPSN, RBCK1, REEP1, RNF216, RRM2B, RTN2, RXYLT1, RYR1, SACS, SBF1, SBF2, SCN10A, SCN11A, SCN4A, SCN9A, SCYL1, SELENON, SEPTIN9, SETX, SGCA, SGCB, SGCD, SGCE, SGCG, SGPL1, SH3TC2, SIGMAR1, SIL1, SLC12A6, SLC1A3, SLC22A5, SLC25A20, SLC25A4, SLC25A42, SLC33A1, SLC52A2, SLC52A3, SLC5A7, SMCHD1, SNAP25, SNX14, 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 myasthene syndromes** (NEM12v22.1; 29 genes)
AGRN, ALG14, ALG2, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COL13A1, COLQ, DOK7, DPAGT1, GFPT1, GMPPB, LAMB2, LRP4, MUSK, MYO9A, PLEC, PREPL, RAPSN, 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, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, GBA2, GJC2, HACE1, HSPD1, IBA57, WASHC5, KIDINS220, KIF1A, KIF1C, KIF5A, KLC2, L1CAM, MAG, MARS2, MTPAP, NIPA1, NT5C2, 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, POGLUT1, POMGNT1, POMT1, POMT2, POPDC3, PTRF, PYROXD1, SGCA, SGCB, SGCD, SGCG, 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

Repeatexpansion 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

☐ **Scapuloperoneal 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, ORAI1, 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/malignt hyperthermia	RYR1
☐	Kennedy disease: X-bound type 1 SBMA, (SMAX1)^	AR^
☐	Motor en sensory neuropathy (d*eletion/duplication test only)*	PMP22/MPZ/GJB1
☐	Myotonic dystrofy type 1 (DM1)^	DMPK^
☐	Myotonic dystrofy type 2 (DM2)^	CNBP^
☐	Spinal Muscular Atrophy (SMA type 1 - 4)$^{\delta}$ *(sequence-analysis only after consultation)*	SMN1$^{\delta}$

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$^{\delta}$ VPS13B$^{\delta}$

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.

$^{\delta}$ 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, NLRC4, NLRP1, NLRP12, 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:
NLRC4, 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, STXBP2, 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, CTNNBL1, 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, GFI1, GINS1, GJC2, GRHL2, GTF2H5, HAVCR2, HAX1, HCK, HELLS, HMOX1, HS3ST6, HYOU1, ICOS, ICOSLG, IFIH1, IFNAR1, IFNAR2, IFNG, IFNGR1, IFNGR2, IGHM, IGLL1, IKBKB, IKBKG, 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, ITPR3, 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, MRTFA, MS4A1, MSN, MTHFD1, MVK, MYD88, MYOF, MYSM1, NBAS, NBN, NCF1, NCF2, NCF4, NCKAP1L, NCSTN, NFAT5, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, NOP10, NOS2, NRAS, NSMCE3, OAS1, ORAI1, OSTM1, OTULIN, PARN, PAX1, PAX5, PBX1, PCCA, PCCB, PDCD1, PEPD, PGM3, PIGA, PIK3CD, PIK3CG, PIK3R1, PLCG2, PLEKHM1, PLG, PMM2, 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, SBDS, 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, STXBP2, SYK, TAP1, TAP2, TAPBP, TAZ, TBX1, TBX21, TCF3, TCIRG1, TCN2, TERT, TET2, TFRC, TGFB1, THBD, TICAM1, TINF2, TIRAP, TLR3, TLR4, TLR7, TLR8, TMC6, TMC8, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF1A, 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, ZNFX1

Primary immunodeficiencies
Single gene | Sequence analysis

☐ VEXAS syndroom 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, ARHGDIA, ARL13B, ARL6, ARMC9, ATXN10, AVIL, B2M, B9D1, B9D2, BBIP1, 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, CSPP1, CTNS, CUBN, CUL3, CYP11B1, CYP11B2, CYP17A1, DAAM2, DACT1, DCDC2, DGKE, DLC1, DNAJB11, DSTYK, E2F3, EMP2, EYA1, FAM149B1, FAN1, FAT1, FGA, FN1, FOXC2, FRAS1, FREM1, FREM2, GANAB, GAPVD1, GATA3, GATM, GLA, GLIS2, GRHPR, GRIP1, GSN, HNF1B, HOGA1, HPSE2, HSD11B2, HYLS1, 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, OCRL, OFD1, OSGEP, PAX2, PBX1, PCM1, PDSS1, PDSS2, PIBF1, PKD1, PKD2, PKHD1, PLCE1, PMM2, POC1B, PODXL, PTPRO, REN, RMND1, ROBO2, RPGRIP1L, RRM2B, SALL1, SARS2, SCARB2, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEC61A1, SGPL1, 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, WDPCP, 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, FOXC1, 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.

[δ] 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, FXYD2, 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, ARHGDIA, 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, CCNQ, 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, CNNM2, COL4A1, COL4A3, COL4A4, COL4A5, COQ2, COQ4, COQ6, COQ7, COQ8B, COQ9, 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, DLC1, DLG5, DMP1, DNAJB11, DOCK4, DST, DSTYK, DYNC2H1, DYNC2LI1, 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, FGFR1, FH, FLCN, FN1, FOXC1, FOXC2, FOXF1, FOXI1, FRAS1, FREM1, FREM2, FXYD2, G6PC, GALNT3, GALT, GANAB, GAPVD1, GATA3, GATM, GCM2, GDF6 , GDNF, GFRA1, GLA, GLI3, GLIS2, GLIS3, GNA11, GNAS, GON7, GPC3, GPC5, GREB1L, GREM1, GRHPR, GRIP1, GSN, GUCY2C, HAAO, HNF1A, HNF1B, HNF4A, HOGA1, HOXA10, HOXA13, HOXD13, HPRT1, HPSE2, HRAS, HSD11B2, HSPA6, HYLS1, 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, PDSS1, PDSS2, PHEX, PIBF1, PKD1, PKD2, PKHD1, PLCE1, PLVAP, PMM2, POC1B, PODXL, PRDM12, PRDX1, PRKCSH, PSAP, PTEN, PTH1R, PTPRO, PYGM, RBM8A, REN, RERE, RET, RICTOR, RMND1, ROBO1, ROBO2, ROR2, RPGRIP1, RPGRIP1L, 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, UQCC2, VDR, VHL, VIPAS39, VPS33B, WBP11, WDPCP, WDR19, WDR34, 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, HPRT1, 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, GRHPR, HNF4A, HOGA1, HPRT1, KCNJ1, KL, 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, ARHGDIA, AVIL, B2M, CD151, CD2AP, CDK20, CFH, CLCN5, COL4A3, COL4A4, COL4A5, COQ2, COQ4, COQ6, COQ7, COQ8B, COQ9, CRB2, CUBN, DAAM2, DGKE, DLC1, 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, PDSS1, PDSS2, 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, DYNC2LI1, DZIP1L, EVC, EVC2, EXOC8, FAM149B1, FAN1, GANAB, GLIS2, GLIS3, GPC3, HNF1B, HYLS1, 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, RMND1, RPGRIP1, RPGRIP1L, SCLT1, SDCCAG8, SEC61A1, SEC61B, SEC63, SLC41A1, SLC4A1, TBX18, TCTEX1D2, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TMEM72, TOGARAM1, TRAF3IP1, TRIM32, TSC1, TSC2, TTC21B, TTC8, TXNDC15, UMOD, VHL, WDPCP, WDR19, WDR34, 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 tubulointerstitual kidney disaese (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, OFD1, PAX2, PKD1, PKD2, PKHD1, 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, RMND1, 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, FGFR1, GALNT3, GATM, GNAS, HRAS, KL, KRAS, NRAS, OCRL, 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, UQCC2, 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.

[δ] 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, ARHGDIA, 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, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, 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, GRHPR, GRIP1, GSN, HNF1B, HOGA1, HOXA13, HPSE2, HSD11B2, HYLS1, IFT122, IFT140, IFT172, IFT27, 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, NCAPG2, NEK1, NEK8, NOS1AP, NOTCH2, NPHP1, NPHP3, NPHP4, 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.

Renal disease
Single gene | Sequence analysis

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

☐ **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** (IPF01v24.1; 28 genes)
ABCA3, ACD, AP3B1, COPA, CSF2RA, CSF2RB, CTC1, DKC1, HPS1, HPS4, NAF1, NHP2, NKX2-1, NOP10, PARN, POT1, RPA1, RTEL1, SFTPA1, SFTPA2, SFTPB, SFTPC, TERC, TERT, TINF2, TMEM173, WRAP53, ZCCHC8

☐ **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, RSPO1, 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, RIPK4, ROR2, RPL10, RSPO1, SAMD9, SEMA3A, SEMA3E, SGPL1, SOHLH1, SOX10, SOX2, SOX3, SOX8, SOX9, SPRY4, SRCAP, SRD5A2, SRY, STAG3, STAR, SYCE1, TAC3, TACR3, TBX3, TOE1, TSPYL1, TWIST2, TWNK, WDR11, WDR60, WNT4, WT1, ZFPM2

Repeat expansion analysis●: ☐ AR

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)[δ]	MYO5B[δ]
☐ Fragile X tremor/ataxia syndrome (FXTAS)^	FMR1^
☐ Premature ovarian falure, (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.

[δ] Sequence and copy number analysis ^ Repeat expansion analysis only

Genome Diagnostics Section
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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.

TESTEN RvA M 001

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**.