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

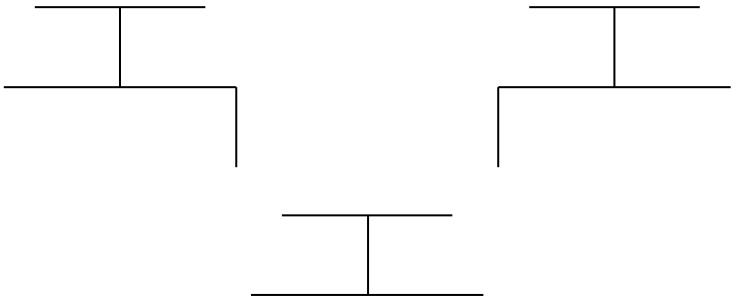
Datum:

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)

Table of contents
Tests available

Blood disorders, vascular disease and bone marrow failure **4**
Gene panels (incl. WES based CNV analysis per gene) 4
Single gene | Sequence analysis 4
Cardiovascular disease **4**
Gene panels (incl. WES based CNV analysis per gene) 4
Single gene | Sequence analysis 5
Dysmorphology **5**
Gene panels (incl. WES based CNV analysis per gene) 5
Single gene | Sequence analysis 5
Epilepsy **5**
Gene panels (incl. WES based CNV analysis per gene) 5
Single gene | Sequence analysis 6
Hereditary cancer **6**
Gene panels (incl. WES based CNV analysis per gene) 6
Single gene | Sequence analysis 6
Intellectual disability: syndromal/non-syndromal **6**
Gene panel | Exome (incl. WES based CNV analysis per gene) 6
Single gene | Sequence analysis 7
Liver diseases **7**
Gene panels (incl. WES based CNV analysis per gene) 7
Metabolic diseases **7**
Gene panels (incl. WES based CNV analysis per gene) 7
Single gene | Sequence analysis 7
Neurological disorders **7**
Gene panels (incl. WES based CNV analysis per gene) 7
Single gene | Sequence / repeat expansion analysis 7
Neuromuscular disease **7**
Gene panels (incl. WES based CNV analysis per gene) 7
Single gene | Sequence analysis 8
Obesity **8**
Gene panels (incl. WES based CNV analysis per gene) 8
Single gene | Sequence analysis 8
Primary immunodeficiencies **9**
Gene panels (incl. WES based CNV analysis per gene) 9
Single gene | Sequence analysis 9
Renal disease **9**
Gene panels (incl. WES based CNV analysis per gene) 9
Single gene | Sequence analysis 11
Other diseases **11**
Gene panels (incl. WES based CNV analysis per gene) 11
Single gene | Sequence analysis 11

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 and/or predisposition for hematologic malignancies** (BMF01v24.2; 252 genes)
ABCB7, ABCD4, ABCG5, ABCG8, ACBD5, ACD, ACKR1, ADA2, AK2, AMN, ANKRD26, AP3B1, ATR, BLM, BRCA1, BRCA2, BRIP1, C15orf41, CA2, CALR, CBL, CD40LG, CEBPA, CLCN7, CLPB, COX4I2, CSF3R, CST1, CTC1, CTLA4, CUBN, CUX1, CXCR4, CYCS, DCLRE1B, DDX41, DHFR, DICER1, DKC1, DNAJC21, DNMT3A, DUT, EFL1, EIF2AK3, ELANE, ENG, ENOSF1, EPCAM, EPG5, ERBB3, ERCC1, ERCC4, ERCC6L2, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FLI1, FYB1, G6PC3, GALE, GATA1, GATA2, GBA, GFI1, GNAS, GNE, GP1BA, GP1BB, GP9, GRHL2, GSKIP, HAX1, HEATR3, HOXA11, HYOU1, IDH1, IDH2, IKZF1, ITGB2, ITGB3, ITK, IVD, JAGN1, JAK2, JAK3, KDM6A, KIT, KLF1, KMT2A, KRAS, LAMTOR1, LIG4, LYST, LZTR1, MAD2L2, MASTL, MBD4, MDM4, MECOM, MEIS1, MLH1, MLPH, MPIG6B, MPL, MRAS, MRTFA, MSH2, MSH6, MTHFD1, MTR, MTRR, MYD88, MYH9, MYSM1, NAF1, NBEAL2, NBN, NF1, NHEJ1, NHP2, NOP10, NOTCH1, NPM1, NRAS, OSTM1, PALB2, PARN, PAX5, PDGFRA, PHF6, PIGA, PLAU, PLCB2, PLEKHM1, PMS2, POT1, PPP1CB, PRF1, PRKACG, PTEN, PTPN11, PUS1, RAB27A, RAC2, RAD21, RAD51C, RAF1, RAP1A, RASGRP2, RBM8A, RFX5, RFXANK, RFXAP, RGS2, RIT1, RMRP, RNF168, RPA1, RPL11, RPL15, RPL26, RPL27A, RPL35A, RPL36, RPL5, RPL8, RPS10, RPS14, RPS15, RPS19, RPS24, RPS26, RPS27A, RPS29, RPS5, RPS7, RPS8, RRAS, RRAS2, RTEL1, RTL1, RUNX1, SAMD9, SAMD9L, SBDS, SBF2, SEC23B, SETBP1, SF3B1, SH2B3, SH2D1A, SHOC2, SLC19A2, SLC25A38, SLC46A1, SLX4, SMARCA3, SMC1A, SMC3, SNX10, SOS1, SOS2, SP1, SRC, SRP54, SRP72, SRSF2, STAG2, STAT3, STAT5A, STEAP3, STIM1, STK4, STN1, TAFAZZIN, TBXAS1, TCIRG1, TCN2, TERC, TERT, TET2, THPO, TINF2, TNFRSF11A, TNFSF11, TP53, TPO, TPP1, TUBB1, TYMS, U2AF1, UBE2T, UBTF, USB1, VPS13B, VPS45, WAS, WRAP53, WT1, XRCC2, YARS2, ZC3HC1, ZCCHC8, ZRSR2

☐ **Diamond-Blackfan anemia** (DBA01v24.1; 41 genes)
EPO, GATA1, HEATR3, RPL10, RPL10A, RPL11, RPL15, RPL17, RPL18, RPL19, RPL26, RPL27, RPL27A, RPL3, RPL31, RPL34, RPL35, RPL35A, RPL36, RPL5, RPL8, RPL9, RPLP0, RPS10, RPS11, RPS14, RPS15, RPS15A, RPS17, RPS19, RPS20, RPS24, RPS26, RPS27, RPS27A, RPS28, RPS29, RPS5, RPS7, RPS8, TSR2

☐ **Hereditary hemolytic anemia** (EMS00v24.1; 48 genes)
ABCB6, ABCG5, ABCG8, ADA, AK1, ALAS2, ALDOA, ANK1, ATP11C, C15orf41, CD59, CDAN1, CYB5R3, EPB41, EPB42, G6PD, GATA1, GCLC, GPI, GPX1, GSR, GSS, HBA1, HBA2, HBB, HBG1, HBG2, HK1, HMOX1, KCNN4, KIF23, KLF1, LCAT, NMNAT3, NT5C3A, PFKM, PGK1, PIEZO1, PKLR, RACGAP1, RHAG, SEC23B, SLC2A1, SLC4A1, SPTA1, SPTB, 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** (EMS01v24.1; 16 genes)
BPGM, CYB5R3, EGLN1, EGLN2, EPAS1, EPO, EPOR, HBA1, HBA2, HBB, JAK2, PIEZO1, PKLR, SH2B3, SLC30A10, VHL

☐ **Rendu Osler Weber syndrome** (ROW01v22.1; 4 genes)
ENG and ACVRL1 copy number analysis included
ENG, ACVRL1, GDF2, SMAD4

Blood disorders and vascular disease
Single gene | Sequence analysis

☐ G6PD deficiency	G6PD
☐ Haemophilia A, (HEMA)[δ]	F8[δ]
☐ Pyruvaat Kinase deficiency (PK)	PKLR
☐ Von Willebrand Factor	VWF

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

☐ **Cardiomyopathy** (CAR01v24.2; 55 genes)
Relevant clinical information
- ☐ *Hypertrophic (HCM)*
- ☐ *Dilated (DCM)^ϞΦ* + ☐ *Conduction abn.*
- ☐ *Arrhythmogenic right ventricle (ARVD/C)*
- ☐ *Left ventricle non compaction (LVNC)*
- ☐ *Restrictive (RCM)*

ACTC1, ACTN2, ALPK3, BAG3, BAG5, CACNA1C, CRYAB, CSRP3, DES, DMD, DSC2, DSG2, DSP, FHL1, FHOD3, FLII, FLNC, GLA, HCN4, JPH2, JUP, KLHL24, LAMP2, LMOD2, LMNA, MIB1, MT-TI, MYBPC3, MYH7, MYL2, MYL3, MYZAP, NEXN, NRAP, PKP2, PLEKHM2, PLN, PPP1R13L, PRDM16, PRKAG2, RBM20, RYR2, SCN5A, TAZ, TCAP, TMEM43, TNNC1, TNNI3, TNNI3K, 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** (CAR05v24.1; 60 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, CHD7, CITED2, CRELD1, ELN, FLT4, FOXH1, GATA4, GATA5, GATA6, GDF1, GJA1, GJA5, GJC1, HAND1, HAND2, HEY1, HEY2, 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, TBX2, TBX20, TBX3, TBX5, TFAP2B, TLL1, ZFPM2, ZIC3

Copy number analysis: ☐ MYBPC3

☐ **Pulmonary Arterial Hypertension (PAH)** (CAR08v24.1; 27 genes)
ABCC8, ACVRL1, AQP1, ATP13A3, BMP10, BMPR1A, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, FBLN2, FOXF1, GDF2, GGCX, KCNK3, KDR, KLF2, KLK1, NOTCH3, PDGFD, SMAD1, SMAD4, 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** (CAR04v24.1; 49 genes)

Relevant clinical information

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

ABL1, ACTA2, ADAMTS19, AEBP1, ARIH1, BGN, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, DCHS1, EFEMP2, ELN, FBN1, FBN2, FLNA, FOXE3, GATA4, GATA5, HCN4, HEY2, IPO8, JAG1, LMOD1, LOX, LTBP3, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, NRP3, PLOD1, PRKG1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, THBS2, THSD4, TLN1

☐ **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, 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
☐ Syndromal microphthalmia 2 (MCOPS2) / Oculofaciocardiodental syndrome (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))

EYA1 copy number analysis included

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)syndromal cleft lip and/or palate incl. Robin sequence** (OWS02v24.1; 203 genes + 1 region (Chr22q11.2))

Requests for this panel are reserved exclusively for clinical geneticists

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 syndrome	ABCC9
☐ Cleidocraniale dysplasia (CCD)[δ]	RUNX2[δ]
☐ Currarino, triad from (TRIAD)	MNX1
☐ Syndromal microphthalmia 2 (MCOPS2) / Oculofaciocardiodental syndrome (OFCD)	BCOR

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

☐ **Epilepsy full gene panel** *(*EPI00v24.1**;** 344 *genes)*

For region chr15:32179526-32273598 copy number analysis included

AARS1, ABAT, ACTL6B, ADPRS, ADSL, AFG2A, ALDH7A1, ALG13, AMT, ANKRD11, ANO4, AP2M1, AP3B2, ARG1, ARHGEF9, ARID1B, ARV1, ARX, ASAH1, ASH1L, ASNS, ASXL3, ATAD1, ATP1A1, ATP1A2, ATP1A3, ATP6AP2, ATP6V0C, ATP6V1A, ATRX, BRAT1, C12orf57, CACNA1A, CACNA1B, CACNA1D, CACNA1E, CACNA1G, CACNA2D2, CAD, CASK, CDK19, CDKL5, CERS1, CHD2, CHD5, CHRNA2, CHRNA4, CHRNB2, CIC, CLCN4, CLDN5, CLN3, CLN5, CLN6, CLN8, CLTC, CNKSR2, CNNM2, CNPY3, CNTNAP2, COQ2, COQ4, CPLX1, CPT2, CSNK2B, CSTB, CTSD, CUL4B, CUX2, CYFIP2, D2HGDH, DCX, DDX3X, DEAF1, DENND5A, DEPDC5, DHDDS, DHPS, DIAPH1, DMXL2, DNAJC5, DNM1, DNM1L, DOCK7, DPM1, DYNC1H1, DYRK1A, EEF1A2, EHMT1, EIF2S3, EIF3F, EPM2A, FARS2, FGD1, FGF12, FGF13, FLNA, FOLR1, FOXG1, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA3, GABRA5, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GAMT, GATM, GCSH, GEMIN5, GLB1, GLDC, GLRA1, GLRB, GNAO1, GNB1, GNB5, GOSR2, GOT2, GPAA1, GPC3, GPHN, GRIA2, GRIA3, GRIA4, GRIK5, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRM7, GRN, HACE1, HCFC1, HCN1, HCN2, HECW2, HNRNPH2, HNRNPR, HNRNPU, HSD17B10, HUWE1, INTS8, IQSEC2, IRF2BPL, ITPA, KANSL1, KAT8, KCNA1, KCNA2, KCNB1, KCNC1, KCNC2, KCND3, KCNH1, KCNH5, KCNJ10, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD3, KCTD7, KDM5C, KIF1A, KIF5C, KMT2A, KPNA7, KPTN, LGI1, LIAS, MAST3, MBD5, MBOAT7, MDH2, MECP2, MED12, MEF2C, MFSD8, MICAL1, MLC1, MOCS1, MOCS2, MPDU1, MTHFR, MTOR, NACC1, NAPB, NBEA, NDE1, NEDD4L, NEU1, NEUROD2, NEXMIF, NHLRC1, NPAP1, NPRL2, NPRL3, NR4A2, NRXN1, NSDHL, NTRK2, NUS1, OFD1, OPHN1, OTUD6B, PACS1, PACS2, PAFAH1B1, PAK1, PAK3, PARS2, PCDH19, PGAP1, PHACTR1, PHF21A, PHF6, PHGDH, PIGA, PIGB, PIGC, PIGG, PIGH, PIGN, PIGO, PIGP, PIGQ, PIGT, PIGU, PIGW, PLCB1, PLP1, PLPBP, PNKP, PNPO, POLG, PPFIBP1, PPP2CA, PPP3CA, PPT1, PQBP1, PRRT2, PSAT1, PSPH, PURA, QARS1, RAB39B, RAI1, RANBP2, RELN, RHOBTB2, RNASEH2A, RNASEH2B, RNASEH2C, ROGDI, RORA, RORB, RPS6KA3, SAMHD1, SCAF4, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SEMA6B, SERPINI1, SETD1A, SETD1B, SHANK3, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC1A2, SLC1A3, SLC1A4, SLC25A1, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SLC6A2, SLC6A8, SLC9A6, SMARCA2, SMC1A, SMS, SNAP25, SON, SPTAN1, ST3GAL3, ST3GAL5, STAMBP, STRADA, STX1B, STXBP1, SYN1, SYNGAP1, SYNJ1, SYP, SZT2, TANC2, TANGO2, TBC1D24, TBCE, TBCK, TBX1, TCF7L2, TDP2, TPP1, TRAK1, TREX1, TRIM8, TRIO, TRPM3, TRPM6, TSC1, TSC2, TUBA1A, TUBB2A, TUBB2B, TUBG1, UBA5, UBE2A, UBE3A, UFM1, UGDH, UGP2, USP25, WDR45, WWOX, YWHAG, ZDHHC9, ZEB2

Repeat expansion analysis●: ☐ CSTB

☐ **Focal epilepsy** (EPI04v24.3; 22 genes)

ATP1A2, CACNA1A, CHRNA2, CHRNA4, CHRNB2, CNKSR2, DCX, DEPDC5, FLNA, GRIN2A, KCNT1, LGI1, MICAL1, MTOR, NPRL2, NPRL3, POLG, RELN, SYN1, TSC1, TSC2, ZDHHC9

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

☐ **Epilepsy (febrile/inflammatory, generalized and/or paroxysmal)** (EPI11v24.1; 34 genes)
For region chr15:32179526-32273598 copy number analysis included
ANO4, ATP1A2, ATP1A3, ATP6V0C, 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, SLC6A1, STX1B, TBC1D24, TBX1, USP25

Deletion/duplication test: ☐ SCN1A ☐ SLC2A1 ☐ PCDH19 ☐ CSTB

Epilepsy
Single gene | Sequence analysis

☐ Dravet syndrome (SMEI/SMEB)[δ]	SCN1A[δ]
☐ Progressive myoclonic epilepsy type 1 / Unverricht Lundborg Disease (ULD) *Repeat expansion analysis included*	CSTB

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

☐ **Breast - and ovarium cancer** (ONC02v22.1; 10 genes)
Requests for this panel are reserved exclusively for clinical geneticists and via mainstreaming procedure
BRCA1 copy number analysis included
BRCA1, BRCA2, ATM, BARD1, CHEK2, PALB2, RAD51C, RAD51D, PTEN, BRIP1

☐ **Ovarian cancer** (ONC01v22.1; 6 genes)
Requests for this panel are reserved exclusively for clinical geneticists and via mainstreaming procedure
BRCA1 copy number analysis included
BRCA1 , BRCA2*, PALB2*, RAD51C, RAD51D, BRIP1 RAD51D*

☐ **Pheochromocytoma** (ONC04v18.1; 11 genes)
SDHAF2, SDHB, SDHC, SDHD 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, SDHB, SDHC and SDHD copy number analysis included
MAX, SDHA, SDHAF2, SDHB, SDHC, SDHD

☐ **Pancreatic cancer** (ONC13v22.1; 6 genes)
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)
AIP, CDKN1B and MEN1 copy number analysis included
AIP, AP2S1, CASR, CDC73, CDKN1A, CDKN1B, CDKN2B, CDKN2C, GNA11, MEN1, RET

☐ **Renal cancer** (ONC07v24.1; 8 genes)
VHL copy number analysis included
BAP1, FH, FLCN, MET, PTEN, SDHB, VHL, PRDM10

☐ **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)
Requests for this panel are reserved exclusively for clinical geneticists
APC and 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)
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)
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)
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

☐ Risk factor for breastcancer / Ataxia-telangiectasia	ATM
☐ Risk factor for 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
☐ Von Hippel-Lindau, disease (VHL)[δ]	VHL[δ]

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

The exome-wide analysis can only be requested by clinical geneticists. Contact us for more information.

☐ **Exome trio analysis intellectual disability** (VBE01v24.1; 1739 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:
Next Generation Sequencing - NGS - UMC Utrecht

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

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]

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

- ☐ **Intrahepatic cholestasis** (HEP01v24.2 (formerly known as MET02); 11 genes)
  ABCB11, ABCB4, ATP8B1, KIF12, LSR, MYO5B, NR1H4, PSKH1, TJP2, USP53, ZFYVE19
- ☐ **Cholestasis, broad differential diagnosis** (HEP02v24.2 (formerly known as MET10); 82 genes)
  ABCB11, ABCB4, ABCC2, ABCD3, ADK, AHCY, AKR1D1, ALDOB, AMACR, ARG1, ASAH1, ATP7B, ATP8B1, BAAT, BCS1L, TWNK, CFTR, UTP4, 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, PSKH1, 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], *deletion test exon 18 included* GAA[δ]
- ☐ 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)
  ATXN2 and C9ORF72 repeat expansion analysis included
  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)
  KRIT1 copy number analysis included
  KRIT1, CCM2, PDCD10
- ☐ **Fahr disease** (NEU04v24.1; 8 genes)
  JAM2, KIAA1161, NAA60, PDGFB, PDGFRB, SLC20A2, XPR1, CMPK2
- ☐ **Sporadic ALS** (NEU05v22.1; 2 genes)
  ATXN2 and C9ORF72 repeat expansion analysis included
  FUS, SOD1
- ☐ **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 / Risk factor for ALS^ ATXN2^

Neuromuscular disease
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.

As of January 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.

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

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)

☐ **Neuropathy** (NMZ01v24.1; 97 genes)
Copy number analysis on NGS data included for all genes in the gene panel (including PMP22/MPZ/GJB1)
AARS1, ABHD12, AIFM1, ARSA, ATL1, ATL3, ATP1A1, ATP7A, BAG3, BICD2, BSCL2, CCT5, COX6A1, CTDP1, DCTN1, DHTKD1, DNAJB2, DNM2, DNMT1, DST, DYNC1H1, EGR2, ELP1, FBLN5, FGD4, FIG4, GAN, GARS1, GBF1, GDAP1, GJB1, GNB4, HARS1, HINT1, HK1, HSPB1, HSPB3, HSPB8, IGHMBP2, INF2, KIF1A, KIF5A, LITAF, LMNA, LRSAM1, MCM3AP, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, NDRG1, NEFH, NEFL, NGF, NTRK1, PDK3, PDXK, PLEKHG5, PMP2, PMP22, PNKP, PRDM12, PRPS1, PRX, RAB7A, REEP1, RETREG1, SBF1, SBF2, SCN10A, SCN11A, SCN9A, SETX, SH3TC2, SIGMAR1, SLC12A6, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SORD, SPG11, SPTLC1, SPTLC2, SURF1, TFG, TRIM2, TRPV4, TTR, VCP, VRK1, VWA1, WARS1, WNK1, YARS1

☐ **Muscle disorders**● (NMZ02v24.1; 215 genes)
To request analysis of the SMN1 (SMA), DMPK (DM1/MD1), or CNBP (DM2/MD2) gene, please submit a separate request under 'Neuromuscular diseases - Single gene.'
ACAD9, ACADVL, ACTA1, ACTN2, ACVR1, ADSS1, AGL, AGRN, ALG14, ALG2, ANO5, ATP2A1, ATP7A, B3GALNT2, B4GAT1, BAG3, BICD2, BIN1, CACNA1S, CAPN3, CASQ1, CAV3, CAVIN1, CFL2, CHAT, CHCHD10, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CLCN1, CLN3, CNTN1, COL12A1, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, CPT2, CRPPA, CRYAB, DAG1, DES, DGUOK, DMD, DNA2, DNAJB4, DNAJB6, DNM2, DNMT3B, DOK7, DPAGT1, DPM1, DPM2, DPM3, DYNC1H1, DYSF, ECEL1, EMD, ENO3, ERBB3, ETFA, ETFB, ETFDH, EXOSC8, FAM111B, FHL1, FKBP14, FKRP, FKTN, FLAD1, FLNC, FXR1, GAA, GATM, GBE1, GFPT1, GMPPB, GNE, GOLGA2, GOSR2, GYG1, GYS1, HACD1, HADHA, HADHB, NRNPA1, HNRNPA2B1, HNRNPDL, HSPB8, HSPG2, IGHMBP2, INPP5K, ISCU, ITGA7, KBTBD13, KLHL40, KLHL41, KLHL9, KY, LAMA2, LAMP2, LARGE1, LDB3, LDHA, LMNA, LMOD3, LPIN1, LRIF1, MAP3K20, MB, MEGF10, MICU1, MSTN, MSTO1, MTM1, MUSK, MYF6, MYH2, MYH3, MYH7, MYL2, MYMK, MYO18B, MYO9A, MYOT, MYPN, NEB, OPA1, ORAI1, PABPN1, PAX7, PFKM, PGAM2, PGK1, PGM1, PHKA1, PIP5K1C, PLEC, PNPLA2, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC1, OPDC3, PREPL, PUS1, PYGM, PYROXD1, RAPSN, RBCK1, RRM2B, RXYLT1, RYR1, SCN4A, SELENON, SGCA, SGCB, SGCD, SGCG, SIL1, SLC18A3, SLC22A5, SLC25A1, SLC25A4, SLC52A2, SLC52A3, SLC5A7, SMCHD1, SMPX, SNUPN, SORD, SPEG, SPTAN1, SPTBN4, SQSTM1, SRPK3, STAC3, STIM1, SUCLA2, SYNE1, SYNE2, SYT2, TANGO2, TCAP, TIA1, TK2, TMEM126B, TMEM43, TNNI2, TNNT1, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM32, TRIP4, TRPV4, TTC19, TTN, TWNK, UBA1, VAMP1, VCP, VIPAS39, VMA21, VRK1, XK, ZC4H2

☐ **Hereditary spastic paraplegia (HSP)** (NMZ03v24.1; 92 genes)
ABCD1, ABHD16A, ADAR, AFG3L2, AIMP1, ALDH18A1, ALDH3A2, ALS2, AMFR, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARG1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BSCL2, C19orf12, CAPN1, COQ4, CPT1C, CYP27A1, CYP2U1, CYP7B1, DDHD1, DDHD2, ENTPD1, ERLIN1, ERLIN2, FA2H, FAR1, FARS2, FBXO7, FXN, GALC, GBA2, GBE1, GCH1, GJA1, GJC2, HACE1, HPDL, HSPD1, IBA57, KCNA2, KIDINS220, KIF1A, KIF1C, KIF5A, KPNA3, L1CAM, MAG, MAPK8IP3, MARS1, MTRFR, NDUFA12, NIPA1, NKX6-2, NT5C2, PCYT2, PLP1, PNPLA6, POLR3A, RAB3GAP2, REEP1, REEP2, RETREG1, RNASEH2B, RNF170, RTN2, SACS, SERAC1, SLC16A2, SLC25A15, SLC2A1, SLC33A1, SPART, SPAST, SPG11, SPG21, SPG7, SPTAN1, TECPR2, TFG, TUBB4A, UBAP1, UCHL1, WASHC5, ZFYVE26

☐ **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)

☐ **Broad NMD panel** (NEM27v24.1; 468 genes)
AARS1, ABCD1, ABHD12, ABHD16A, ACAD9, ACADVL, ACTA1, ACTN2, ACVR1, ADAR, ADSS1, AFG3L2, AGL, AGRN, AIFM1, AIMP1, ALDH18A1, ALDH3A2, ALG14, ALG2, ALS2, AMFR, AMPD2, ANO10, ANO5, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APTX, AR, ARG1, ARL6IP1, ARSA, ASAH1, ASCC1, ATG7, ATL1, ATL3, ATM, ATP13A2, ATP1A1, ATP2A1, ATP7A, B3GALNT2, B4GALNT1, B4GAT1, BAG3, BEAN1, BICD2, BIN1, BSCL2, C19orf12, CACNA1A, CACNA1G, CACNA1S, CACNB4, CAPN1, CAPN3, CASQ1, CAV3, CAVIN1, CCDC78, CCDC88C, CCT5, CFL2, CHAT, CHCHD10, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CLCN1, CLN3, CNTN1, CNTNAP1, COA7, COL12A1, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, COQ4, COQ8A, COX6A1, CPT1C, CPT2, CRPPA, CRYAB, CTDP1, CWF19L1, CYP27A1, CYP2U1, CYP7B1, DAG1, DCTN1, DDHD1, DDHD2, DES, DGUOK, DHTKD1, DMD, DNA2, DNAJB2, DNAJB4, DNAJB6, DNM2, DNMT1, DNMT3B, 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, FAR1, FARS2, FASTKD2, FBLN5, FBXO38, FBXO7, FGD4, FGF14, FHL1, FIG4, FKBP14, FKRP, FKTN, FLAD1, FLNC, FXN, FXR1, GAA, GALC, GAN, GARS1, GATM, GBA2, GBE1, GBF1, GCH1, GDAP1, GDAP2, GFPT1, GJA1, GJB1, GJC2, GLE1, GMPPB, GNB4, GNE, GOLGA2, GOSR2, GRID2, GRM1, GYG1, GYS1, HACD1, HACE1, HADHA, HADHB, HARS1, HEXB, HINT1, HK1, HNRNPA1, HNRNPA2B1, HNRNPDL, HPDL, HSPB1, HSPB3, HSPB8, HSPD1, HSPG2, IBA57, IFRD1, IGHMBP2, INF2, INPP5K, ISCU, ITGA7, ITPR1, KARS1, KBTBD13, KCNA1, KCNA2, KCNC3, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNJ18, KCNJ2, KCNJ5, KIDINS220, KIF1A, KIF1C, KIF21A, KIF5A, KLHL40, KLHL41, KLHL9, KPNA3, KY, L1CAM, LAMA2, LAMB2, LAMP2, LARGE1, LDB3, LDHA, LITAF, LMNA, LMOD3, LPIN1, LRIF1, LRSAM1, MAG, MAP3K20, MAPK8IP3, MARS1, MARS2, MB, MCM3AP, MED25, MEGF10, MFN2, MICU1, MME, MORC2, MPV17, MPZ, MRE11, MSTN, MSTO1, MTM1, MTMR2, MTPAP, MTRFR, MUSK, MYF6, MYH2, MYH3, MYH7, MYH8, MYL2, MYMK, MYO18B, MYO9A, MYOT, MYPN, NDRG1, NDUFA12, NEB, NEFH, NEFL, NGF, NIPA1, NKX6-2, NOP56, NT5C2, NTRK1, OBSCN, OPA1, ORAI1, PABPN1, PAX7, PCYT2, PDK3, PDXK, PDYN, PEX7, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHOX2A, PHYH, PIP5K1C, PLEC, PLEKHG5, PLP1, PMP2, PMP22, PNKP, PNPLA2, PNPLA6, POGLUT1, POLG, POLG2, POLR3A, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC1, POPDC3, PRDM12, PREPL, PRKCG, PRPS1, PRX, PTRH2, PUS1, PYGM, PYROXD1, RAB3GAP2, RAB7A, RAPSN, RBCK1, REEP1, REEP2, RETREG1, RNASEH2B, RNF170, RNF216, RRM2B, RTN2, RXYLT1, RYR1, SACS, SBF1, SBF2, SCN10A, SCN11A, SCN4A, SCN9A, SCYL1, SELENON, SEPTIN9, SERAC1, SETX, SGCA, SGCB, SGCD, SGCE, SGCG, SGPL1, SH3TC2, SIGMAR1, SIL1, SLC12A6, SLC16A2, SLC18A3, SLC1A3, SLC22A5, SLC25A1, SLC25A15, SLC25A4, SLC25A42, SLC25A46, SLC2A1, SLC33A1, SLC52A2, SLC52A3, SLC5A7, SMCHD1, SMPX, SNUPN, SNX14, SORD, SPART, SPAST, SPEG, SPG11, SPG21, SPG7, SPTAN1, SPTBN2, SPTBN4, SPTLC1, SPTLC2, SQSTM1, SRPK3, STAC3, STIM1, STUB1, SUCLA2, SURF1, SYNE1, SYNE2, SYT2, TANGO2, TCAP, TDP1, TDP2, TECPR2, TFG, TGM6, THG1L, TIA1, TK2, TMEM126B, 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, TUBB4A, TWNK, UBA1, UBAP1, UCHL1, VAMP1, VCP, VIPAS39, VMA21, VPS13D, VPS37A, VRK1, VWA1, WARS1, WASHC5, WNK1, XK, YARS1, YARS2, ZC4H2, ZFYVE26, ZFYVE27

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

Neuromuscular diseases
Single gene | Sequence analysis

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

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[δ]	VPS13B[δ]

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

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** (PID00v24.1; 479 genes)
ACD, ACP5, ACTB, ADA, ADA2, ADAM17, ADAR, AGA, AICDA, AIRE, AK2, ALG13, ALPI, ALPK1, ANGPT1, AP1S3, AP3B1, AP3D1, APOL1, ARHGEF1, ARPC1B, ARPC5, 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, 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, DPP9, ELANE, ELF4, EPG5, ERBIN, ERCC6L2, EXTL3, F12, FAAP24, FADD, FAS, FASLG, FAT4, FCGR3A, FCHO1, FCN3, FERMT1, FERMT3, FNIP1, FOXI3, FOXN1, FOXP3, FPR1, G6PC, G6PC3, G6PD, GATA1, 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, IL1R1, IL1RN, IL2, IL21, IL21R, IL2RA, IL2RB, IL2RG, IL36RN, IL6R, IL6ST, IL7R, INO80, INSR, IRAK1, IRAK4, IRF1, 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, LYN, MAGT1, MALT1, MAN2B1, MAN2B2, MANBA, MAP1LC3B2, MAP3K14, MAPK8, MASP2, MC2R, MCM10, MCM4, MCTS1, 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, PBX1, PCCA, PCCB, PDCD1, PEPD, PGM3, PI4KA, PIGA, PIK3CD, PIK3CG, PIK3R1, PLCG2, PLEKHM1, PLG, PMM2, PNP, POLA1, POLE2, POLR3F, POMP, POT1, POU2AF1, PRF1, PRKCD, PRKDC, PRPS1, PSENEN, PSMA3, PSMB4, PSMB8, PSMB9, PSMB10, PSMG2, PSTPIP1, PTEN, PTPN22, PTPRC, RAB27A, RAC2, RAG1, RAG2, RANBP2, RASGRP1, RASGRP2, RBCK1, RC3H1, RECQL4, REL, RELA, RELB, RFX5, RFXANK, RFXAP, RHOG, RHOH, RIPK1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RNU4ATAC, RNU7-1, 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, SNORA31, 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, TERC, 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 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)
CD46, CFH, CFI, CFHR1 and CFHR3 copy number analysis included
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)** (NEF24v24.1; 263 genes)
ACE, ACTG2, ACTN4, ADAMTS9, ADAMTS13, 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, PRDX1, 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, THBD, 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, ZNF4
In general the analysis will not detect MUC1 VNTR Cytosine-insertions.
Copy number analysis: ☐ HNF1B ☐ NPHP1

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)

☐ **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, 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)
Requests for this panel are reserved exclusively for clinical geneticists; please use NEF24 in case of kidney failure of unknown cause
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
In general the analysis will not detect MUC1 VNTR Cytosine-insertions.

☐ **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

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 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, 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** (NEF18v24.1; 34 genes)
ALDOB, ALPL, AP2S1, CASR, CLCN5, CTNS, CYP27B1, CYP2R1, CYP3A4, DMP1, EHHADH, ENPP1, FAH, FAM20C, FGF23, FGFR1, G6PC, GALNT3, GALT, GATM, GNA11, GNAS, HRAS, KL, KRAS, NRAS, OCRL, PHEX, PTH1R, SLC2A2, 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

☐ **Renal Tubular Dysgenesis** (NEF20v16.1; 5 genes)
ACE, AGT, AGTR1, REN, UMOD

☐ **Chronic kidney disease-kids, CKD-kids *(including PKD1 and PKD2)*** (NEF27v24.1; 363 genes)
ACE, ACTG2, ACTN4, ADAMTS13, 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, PRDX1, 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, THBD, 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 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** (AMY01v24.1; 15 genes)
APOA1, APOA2, APOC2, APOC3, APP, B2M, CST3, FGA, GSN, IL31RA, ITM2B, LYZ, NLRP3, 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)
SRY, SOX9, NR0B1 and SOX3 copy number analysis included
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)
SRY, SOX9, NR0B1 and SOX3 copy number analysis included
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
Department of Genetics
University Medical Center (UMC) Utrecht
Heidelberglaan 100
3584 CX Utrecht

UMC 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 Use of residual material - UMC Utrecht)

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**.