SEND TO
Genome Diagnostics Section
University Medical Center Utrecht
Centrale Balie LKCH
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
Fax +31 (0)88 – 75 55034
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
☐ Diagnostic testing
☐ 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) : .....
☐ 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

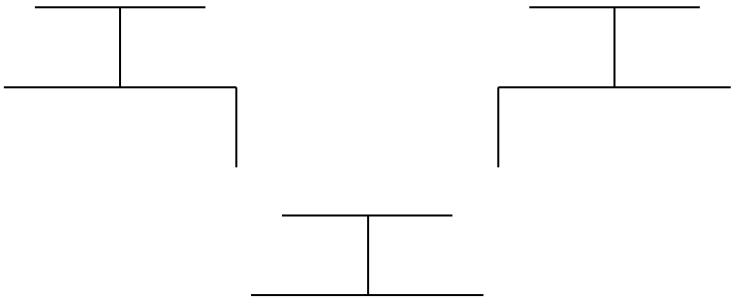
Datum:

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

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 and vascular disease
Gene panels

☐ **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 haemostasis** (TRO02v17.1; 90 genes)
ABCG5, ABCG8, ACTN1, ACVRL1, ADRA2A, ADRA2B, ANKRD26, ANO6, AP3B1, BLOC1S3, BLOC1S6, CD36, CDC42, COL1A1, COL5A1, COL5A2, COL3A1, CYCS, DTNBP1, ENG, ETV6, F2R, F2RL3, FBN1, FERMT3, FGA, FGB, FGG, FLI1, FLNA, FYB, GATA1, GATA2, GBA, GFI1B, GNAI1, GNAI2, GNA12, GNA13, GNAZ, GNAS, GNAQ, GNE, GP1BA, GP1BB, GP6, GP9, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA2, ITGA2B, ITGB1, ITGB3, LYST, MASTL, MECOM, MLPH, MPL, MYH9, MYO5A, NBEAL2, P2RX1, P2RY1, P2RY12, PLA2G4A, PLAU, PLCB2, PLCB3, PLCG2, PRKACG, PTGS1, RAB27A, RASGRP2, RBM8A, RGS2, RUNX1, SLFN14, STIM1, TBXA2R, TBXAS1, THPO, TUBB1, VPS33B, VIPAS39, VWF, WAS

☐ **Congenital secondary erythrocytosis** (EMS01v20.1; 15 genes)
EPOR, VHL, EGLN1, EPAS1, EPO, HBB, HBA1, HBA2, BPGM, PKLR, PIEZO1, SH2B3, EGLN2, HIF3A, OS9

Blood disorders and vascular disease
Single gene | Sequence analysis

☐ Haemophilia A, (HEMA)[δ]	F8[δ]
☐ Hereditary haemorrhagic telangiectasia 1 (HHT1) / Rendu-Osler-Weber syndrome (ROW)[δ]	ENG[δ]
☐ Hereditary haemorrhagic telangiectasia 2 (HHT2) / Rendu-Osler-Weber syndrome (ROW)[δ]	ACVRL1[δ]
☐ Hereditary haemorrhagic telangiectasia 5 (HHT5) / Rendu-Osler-Weber disease (ROW)	GDF2
☐ Juvenile polyposis / Hereditary haemorrhagic telangiectasia syndrome (JPHT)	SMAD4
☐ Thrombocythemia 1	THPO
☐ Thrombocytopenia, congenital amegakaryocytic (CAMT)	MPL
☐ Von Willebrand Factor *[TRO03v18.1]*	VWF

Cardiovascular disease
Gene panels

☐ **Cardiomyopathy*** (CAR01v16.1; 64 genes)
Relevant clinical information
- ☐ *Hypertrophic (HCM)*
- ☐ *Dilated (DCM)*[∞] + ☐ *Conduction abn.*
- ☐ *Arrhythmogenic right ventricle (ARVD/C)*
- ☐ *Left ventricle non compaction (LVNC)*
- ☐ *Restrictive (RCM)*

ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, CALR3, CASQ2, CAV3, CRYAB, CSRP3, CTNNA3, DES, DMD, DSC2, DSG2, DSP, DTNA, EMD, EYA4, FHL1, FLNC, FKTN, GATAD1, GLA, ILK, JPH2, JUP, LAMA4, LAMP2, LDB3, LMNA, MIB1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOT, MYOZ1, MYOZ2, MYPN, NEBL, NEXN, PDLIM3, PKP2, PLN, PRKAG2, RBM20, RYR2, SCN5A,SGCD, TAZ, TCAP, TGFB3, TMEM43, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTR, VCL

Copy number analysis*: ☐ MYBPC3 ☐ PKP2

[∞] Titin gene mutations are found to underlie a substantial part of dilated cardiomyopathy (DCM) cases and must be requested separately (see below).

☐ **Conduction abnormalities*** (CAR03v18.1; 37 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)*

AKAP9, ANK2, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, DES, DPP6, DSC2, DSG2, DSP, GPD1L, HCN4, JUP, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, LMNA, PKP2, PLN, RYR2, SCN1B, SCN3B, SCN4B, SCN5A, SNTA1, TGFB3, TMEM43

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 ☐ JAG1

☐ **Vascular disorders** (CAR04v20.1; 39 genes)
Relevant clinical information
- ☐ *Familial thoracic aortic aneurysm and aortic dissection (TAAD)*
- ☐ *Marfan (MFS)*
- ☐ *Loeys-Dietz (LDS)*

ACTA2, BGN, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, DCHS1, EFEMP2, ELN, EMILIN1, FBN1, FBN2, FLNA, FOXE3, GATA4, GATA5, HCN4, LMOD1, LOX, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, ROBO4, SCARF2, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2

Cardiovascular disease
Single gene | Sequence analysis

☐ Alagille syndrome *(copy number analysis only)*	JAG1
☐ Alveolar capillary dysplasia with misalignment of the pulmonary veins (ACDMPV)	FOXF1
☐ AR right atrium isomerism	GDF1
☐ Arrhythmogenic right ventricular dysplasia (ARVD/C1)	TGFB3
☐ Arrhythmogenic right ventricular dysplasia (ARVD/C5)	TMEM43
☐ Arrhythmogenic right ventricular dysplasia (ARVD/C8)	DSP
☐ Arrhythmogenic right ventricular dysplasia (ARVD/C9)[δ]	PKP2[δ]
☐ Arrhythmogenic right ventricular dysplasia (ARVD/C10)	DSG2
☐ Arrhythmogenic right ventricular dysplasia (ARVD/C11)	DSC2
☐ Arrhythmogenic right ventricular dysplasia (ARVD/C12)	JUP
☐ Arrhythmogenic right ventricular dysplasia (ARVD/C)	DES
☐ Arrhythmogenic right ventricular dysplasia (ARVD/C)	PLN

* NGS gene panel analysis can only detect single nucleotide changes and small deletions/duplications. Large copy number changes and repeat expansions cannot be detected. Unless indicated otherwise, these analyses must be requested seperately.

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

Cardiovascular disease (Continued)
Single gene | Sequence analysis

☐ Arrhythmogenic right ventricular dysplasia (ARVD/C)[δ]	LMNA[δ]
☐ Arrhythmogenic Right Ventricular Dysplasia/ cardiomyopathy (ARVD/C)	CTNNA3
☐ Brugada syndrome	SCN1B
☐ Cantú syndrome	ABCC9
☐ Cardiomyopathy, dilated (DCM)[δ]	LMNA[δ]
☐ Cardiomyopathy, dilated (DCM)	DES
☐ Cardiomyopathy, dilated (DCM), Titin gene analysis *[CAR06v16.1]*	TTN
☐ Cardiomyopathy, dilated and cataract (DCM)	CRYAB
☐ Cardiomyopathy, dilated, hypertrophic (DCM/HCM)	TNNT2
☐ Cardiomyopathy, dilated, hypertrophic (DCM/HCM)	PLN
☐ Cardiomyopathy, dilated, hypertrophic (DCM/HCM)	MYL2
☐ Cardiomyopathy, dilated, hypertrophic (DCM/HCM)	MYLK2
☐ Cardiomyopathy, dilated, hypertrophic (DCM/HCM)	MYOZ2
☐ Cardiomyopathy, dilated, hypertrophic (DCM/HCM)	MYH7
☐ Cardiomyopathy, dilated, hypertrophic (DCM/HCM)[δ]	MYBPC3[δ]
☐ Cardiomyopathy, dilated, hypertrophic (DCM/HCM)	CASQ2
☐ Cardiomyopathy, dilated, hypertrophic (DCM/HCM)	CAV3
☐ Cardiomyopathy, dilated, hypertrophic (DCM/HCM)	FHL1
☐ Cardiomyopathy, dilated, hypertrophic (DCM/HCM)	TCAP
☐ Cardiomyopathy, dilated, hypertrophic (DCM/HCM)	TNNC1
☐ Cardiomyopathy, dilated, hypertrophic (DCM/HCM)	TNNI3
☐ Cardiomyopathy, dilated, hypertrophic (DCM/HCM)	TPM1
☐ Cataract and dilated cardiomyopathy	CRYAB
☐ Fabry disease, alpha-galactosidase A deficiency[δ]	GLA[δ]
☐ Fallot, Tetralogy of (TOF)	NKX2-5
☐ Fallot, Tetralogy of (TOF), AD	GDF1
☐ Holt-Oram syndrome (HOS)[δ]	TBX5[δ]
☐ Long QT syndrome, type 1 and 2 *(copy number analysis only)*	KCNQ1/KCNH2
☐ Oculofaciocardiodental syndrome (OFCD)	BCOR
☐ Syndromal microphthalmia 2 (MCOPS2)	BCOR
☐ Velocardiofacial syndrome (VCF) / DiGeorge Syndrome	TBX1
☐ Ventricular tachycardia, catecholaminergic polymorphic type 2 (CPVT2)	CASQ2

Dysmorphology
Gene panels

☐ **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

☐ **Fraser syndrome** (FRA00v16.1; 4 genes)
FRAS1, FREM2, FREM1, GRIP1

☐ **Hemifacial microsomia** (OWS01v19.1; 43 genes)
Includes copy number analysis of EYA1
BMP4, CDC6, CDT1, CHD7, DHODH, EDNRA, EFTUD2, EIF4A3, EYA1, FGF10, FGF3, FGFR2, FGFR3, FRAS1, FREM2, GNAI3, GRIP1, GSC, HMX1, HOXA2, HSPA9, KDM6A, KMT2D, OFD1, ORC1, ORC4, ORC6, OTX2, PLCB4, POLR1A, POLR1C, POLR1D, SALL1, SALL4, SF3B4, SIX1, SLC26A4, SOX10, TCOF1, TFAP2A, GDF6, RPS28, SIX5

☐ **Hypodontia/Oligodontia** (DON01v19.1; 17 genes)
AXIN2, BCOR, EDA, EDAR, EDARADD, FGFR1, FLNA, GJA1, GREM2, IRF6, LRP6, LTBP3, MSX1, PAX9, TP63, WNT10A, WNT10B

☐ **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

☐ **(Non)syndromaal cleft lip and/or palate incl. Robin sequence** (OWS02v20.2; 195 genes)
Pre-test genetic counselling required
ACTB, ACTG1, ALX1, ALX3, AMER1, AMMECR1, ANKRD11, ARHGAP29, ARHGAP31, ASXL1, B3GALT6, B3GALTL, B4GALT7, B9D2, BCOR, BMP2, BMPER, C2CD3, C5orf42, CC2D2A, CDC45, CDH1, CDKN1C, CHD7, CHRNG, CHST14, COL11A1, COL11A2, COL2A1, COL9A1, COLEC10, COLEC11, CTCF, CTNND1, DDX3X, DDX59, DHCR7, DHODH, DLL4, DOCK6, DVL1, DVL3, DYNC2H1, 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, 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, SEPT9, SF3B4, SHH, SIX1, SIX3, SIX5, SKI, SLC26A2, SMAD3, SMAD4, SMC1A, SMC3, SMCHD1, SMS, SNRPB, SON, SOX9, SPECC1L, STAC3, STAMBP, TAPT1, TBX1, TBX15, TBX2, TBX22, TCOF1, TCTN3, TFAP2A, TGDS, TGFB3, TGFBR1, TGFBR2, TGIF1, TMCO1, TMEM216, TMEM8C, TP63, TRIM37, TUBB, TWIST1, TXNL4A, USP9X, WDR35, WNT4, WNT5A, XYLT1, ZEB2, ZIC2, ZIC3, ZMPSTE24, ZSWIM6

Dysmorphology
Single gene | Sequence analysis

☐ Acrocallosaal Syndrome (ACLS)	KIF7
☐ Albright hereditary osteodystrophy (AHO) *(sequence-analysis and methylation specific copy number analysis)*	GNAS
☐ Amelogenesis imperfecta, hypomaturation-hypoplasic type, with taurodontism (AIHHT)	DLX3
☐ Cantú syndrome	ABCC9
☐ Cleidocranial dysplasia (CCD)[δ]	RUNX2[δ]
☐ Currarino syndrome, TRIAD	MNX1
☐ Floating-Harbor Syndrome (FHS)	SRCAP
☐ Hypodontia (HYD1)	MSX1
☐ Hypodontia (HYD3)	PAX9
☐ Hypodontia	WNT10A
☐ Hypodontia / Oligodontia	IRF6
☐ Hypodontia / Oligodontia	ITM2A
☐ Hypodontia / Oligodontia	SUMO1
☐ Hypodontia / Oligodontia	TBX22
☐ Hypodontia / Oligodontia-colorectal cancer syndrome (ODCRCS)	AXIN2
☐ McCune-Albright syndrome, (MAS) / Osseous heteroplasia progressive, (POH)	GNAS
☐ Microphthalmia, syndromic 2 (MCOPS2) / Oculofaciocardiodental syndrome (OFCD)	BCOR
☐ Pseudohypoparathyroidism, type 1A (PHP1A)[δ] *(sequence-analysis and methylation specific copy number analysis)*	GNAS
☐ Trichodontoosseous syndrome (TDO)	DLX3
☐ Van der Woude syndrome	IRF6

Epilepsy
Gene panels

☐ **IGE/JME/CAE*** (EPI07v18.1; 7 genes)
CACNB4, CHD2, GABRA1, GABRB3, SCN8A, SLC2A1, SLC6A1
Copy number analysis*: ☐ SLC2A1

☐ **Progressive myoclonic epilepsy*** (EPI05v18.1; 14 genes)
ASAH1, CERS1, CSNK2B, EPM2A, GOSR2, IRF2BPL, KCNA2, KCNC1, KCTD7, NHLRC1, POLG, PRICKLE1, PRICKLE2, SCARB2
Copy number analysis*: ☐ EPM2A ☐ NHLRC1

* NGS gene panel analysis can only detect single nucleotide changes and small deletions/duplications. Large copy number changes and repeat expansions cannot be detected. Unless indicated otherwise, these analyses must be requested seperately.

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

Epilepsy (Continued)
Gene panels

☐ **Epilepsy full gene panel** (EPI00v18.1; 200 *genes)*
AARS, ACTL6B, ADSL, ALDH7A1, ALG13, AMT, ANKRD11, AP3B2, ARHGEF9, ARV1, ARX, ASAH1, ATAD1, ATP1A2, ATP1A3, ATP6AP2, ATRX, BRAT1, CACNA1A, CACNB4, CASK, CDKL5, CERS1, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN4, CLN3, CLN5, CLN6, CLN8, CNKSR2, CNTNAP2, COQ4, CPT2, CSNK2B, CTNND2, CTSD, CUL4B, DCX, DENND5A, DEPDC5, DNAJC5, DNM1, DOCK7, DYRK1A, EEF1A2, EPM2A, FGD1, FLNA, FOLR1, FOXG1, FRRS1L, GABRA1, GABRA3, GABRB3, GABRG2, GAMT, GCSH, GLDC, GLRA1, GLRB, GNAO1, GOSR2, GPC3, GPHN, GRIA3, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRN, HCFC1, HCN1, HNRNPU, HSD17B10, HUWE1, INTS8, IQSEC2, IRF2BPL, KCNA2, KCNB1, KCNC1, KCND3, KCNH1, KCNJ10, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCTD7, KDM5C, KIAA2022, KMT2A, KPNA7, LGI1, MBD5, MDH2, MECP2, MED12, MEF2C, MFSD8, MOCS1, MOCS2, MTHFR, mTOR, NAPB, NBEA, NHLRC1, NPRL2, NPRL3, NRXN1, NSDHL, OFD1, OPHN1, PAK3, PCDH19, PGAP1, PHF6, PHGDH, PIGA, PIGN, PIGO, PIGT, PLCB1, PLP1, PNKP, PNPO, POLG, PPP3CA, PPT1, PQBP1, PRICKLE1, PRICKLE2, PRIMA1, PRRT2, PSAT1, PSPH, PURA, QARS, RAB39B, RAI1, RANBP2, RELN, RNASEH2A, RNASEH2B, RNASEH2C, ROGDI, RPS6KA3, SAMHD1, SCARB2, SCN1A, SCN1B, SCN2A, SCN8A, SHANK3, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC1A3, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SLC6A5, SLC6A8, SLC9A6, SMC1A, SMS, SNAP25, SON, SPTAN1, ST3GAL3, STX1B, STXBP1, SYN1, SYNGAP1, SYNJ1, SYP, SZT2, TBC1D24, TBCE, TBCK, TCF4, TPP1, TREX1, TRIO, UBA5, UBE2A, UBE3A, UGDH, WDR45, WWOX, YWHAG, ZDHHC9, ZEB2

☐ **Epileptic encephalopathy (EIEE)*** (EPI02v18.1; 90 genes)
ANKRD11, AP3B2, FRRS1L, KCNB1, UBA5, WWOX, ACTL6B, ALDH7A1, ALG13, ARHGEF9, ARV1, ARX, ATAD1, ATP1A3, BRAT1, CDKL5, CHD2, CNKSR2, CSNK2B, DENND5A, DEPDC5, DNM1, DOCK7, EEF1A2, FOXG1, GABRA1, GABRA3, GABRB3, GNAO1, GRIN1, GRIN2A, GRIN2B, GRIN2D, HCFC1, HCN1, HNRNPU, HUWE1, IRF2BPL, KCNA2, KCNQ2, KCNQ3, KCNQ5, KCNT1, KIAA2022, KPNA7, MDH2, MECP2, MEF2C, MOCS1, MOCS2, NAPB, NBEA, PCDH19, PHGDH, PLCB1, PNKP, PNPO, POLG, PRRT2, PSAT1, PSPH, PURA, SCN1A, SCN1B, SCN2A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SNAP25, SPTAN1, ST3GAL3, STX1B, STXBP1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TBCE, TRIO, UBE3A, UGDH, WDR45, YWHAG, ZEB2
Copy number analysis*: ☐ ARX ☐ CDKL5 ☐ FOXG1 ☐ KCNQ2 ☐ MECP2 ☐ MEF2C ☐ PCDH19 ☐ SCN1A ☐ SLC2A1

☐ **Febrile seizures / Genetic epilepsy with febrile seizures plus (GEFS+)** (EPI03v18.1; 173 genes)
ATP1A2, CACNA1A, CHD2, CLCN4, GABRA1, GABRB3, GABRG2, HCN1, KCNA2, PCDH19, POLG, SCN1A, SCN1B, SCN2A, SCN8A, STX1B, TBC1D24
Copy number analysis*: ☐ PCDH19 ☐ SCN1A

☐ **Focal epilepsy*** (EPI04v18.1; 19 genes)
CHRNA2, CHRNA4, CHRNB2, CNKSR2, DCX, DEPDC5, FLNA, GRIN2A, KCNT1, LGI1, mTOR, NPRL2, NPRL3, POLG, PRIMA1, RELN, SLC12A5, SYN1, ZDHHC9
Copy number analysis*: ☐ CHRNA4 ☐ CHRNB2

☐ **Metabolic disease with epilepsy*** (EPI06v18.1; 38 genes)
ADSL, ALDH7A1, ALG13, AMT, CLN3, CLN5, CLN6, CLN8, CPT2, CTSD, DNAJC5, FOLR1, GAMT, GCSH, GLDC, GLRA1, GLRB, GPHN, GRN, HCFC1, MDH2, MFSD8, MOCS1, MOCS2, MTHFR, PHGDH, PIGA, PIGN, PIGT, PNPO, POLG, PPT1, PSAT1, PSPH, SLC2A1, SLC35A2, SLC6A8, TPP1
Copy number analysis*: ☐ GLDC ☐ SLC2A1

☐ **Epileptic syndromes with epilepsy and intellectual disability*** (EPI09v18.1; 117 genes)
ANKRD11, ALG13, AARS, AP3B2, FRRS1L, KCNB1, UBA5, WWOX, ACTL6B, ARV1, ARX, ATAD1, ATP1A3, ATP6AP2, ATRX, CASK, CDKL5, CHD2, CLCN4, CNKSR2, CNTNAP2, COQ4, CSNK2B, CUL4B, DCX, DENND5A, DOCK7, DYRK1A, EEF1A2, FGD1, FLNA, FOXG1, GABRA3, GPC3, GRIA3, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, HCFC1, HNRNPU, HSD17B10, HUWE1, INTS8, IQSEC2, IRF2BPL, KCNA2, KCND3, KCNH1, KCNJ10, KCNQ5, KDM5C, KIAA2022, KMT2A, KPNA7, MBD5, MDH2, MECP2, MED12, MEF2C, NAPB, NBEA, NRXN1, NSDHL, OFD1, OPHN1, PAK3, PGAP1, PHF6, PIGA, PIGN, PIGO, PIGT, PLP1, PNKP, POLG, PPP3CA, PQBP1, PURA, QARS, RAB39B, RAI1, RNASEH2A, RNASEH2B, RNASEH2C, ROGDI, RPS6KA3, SAMHD1, SCN8A, SHANK3, SLC13A5, SLC35A2, SLC6A1, SLC6A8, SLC9A6, SMC1A, SMS, SNAP25, SON, ST3GAL3, STXBP1, SYNGAP1, SYP, SZT2, TBC1D24, TBCK, TCF4, TREX1, TRIO, UBE2A, UBE3A, UGDH, WDR45, YWHAG, ZDHHC9, ZEB2
Copy number analysis*: ☐ ARX ☐ CDKL5 ☐ FOXG1 ☐ MECP2 ☐ MEF2C ☐ NRXN1

☐ **Inflammatory epilepsy*** (EPI10v17.1; 3 genes)
CPT2, RANBP2, SCN1A
Copy number analysis*: ☐ SCN1A

☐ **Epilepsy with paroxysmal disorders*** (EPI08v18.1; 11 genes)
ATP1A2, ATP1A3, CACNA1A, KCNA2, KCNMA1, PRRT2, SCN1A, SCN8A, SLC1A3, SLC2A1, CTNND2
Copy number analysis*: ☐ SLC2A1

Epilepsy
Single gene | Sequence analysis

☐ Autosomal dominant lateral temporal lobe epilepsy (ADLTE)	LGI1
☐ Benign familial infantile seizures type 2 (BFIS2)	PRRT2
☐ Benign familial neonatal seizures (BFNC)[δ]	KCNQ2[δ]
☐ Benign familial neonatal seizures (BFNC)[δ]	KCNQ3[δ]
☐ Benign familial neonatal-infantile seizures (BFNIS)	SCN2A
☐ Cortical dysplasia-focal epilepsy syndrome (CDFE)	CNTNAP2
☐ Dravet syndrome (SMEI/SMEB)[δ]	SCN1A[δ]
☐ Early infantile epileptic encephalopathy type 1 (EIEE1)[δ]	ARX[δ]
☐ Early infantile epileptic encephalopathy type 2 (EIEE2)[δ]	CDKL5[δ]
☐ Early infantile epileptic encephalopathy type 3 (EIEE3)	SLC25A22
☐ Early infantile epileptic encephalopathy type 4 (EIEE4)[δ]	STXBP1[δ]
☐ Early infantile epileptic encephalopathy type 7 (EIEE7)[δ]	KCNQ2[δ]
☐ Early infantile epileptic encephalopathy type 8 (EIEE8)	ARHGEF9
☐ Early infantile epileptic encephalopathy type 9 (EIEE9)[δ]	PCDH19[δ]
☐ Early infantile epileptic encephalopathy type 10 (EIEE10)	PNKP
☐ Early infantile epileptic encephalopathy type 11 (EIEE11)	SCN2A
☐ Early infantile epileptic encephalopathy type 12 (EIEE12)	PLCB1
☐ Genetic epilepsy with febrile seizures plus (GEFS+)[δ]	SCN1A[δ]
☐ Genetic epilepsy with febrile seizures plus (GEFS+)	SCN1B
☐ Genetic epilepsy with febrile seizures plus (GEFS+)	SCN2A
☐ Genetic epilepsy with febrile seizures plus (GEFS+)	GABRG2
☐ GLUT1 deficiency syndrome type 1 and 2, (GLUT1DS1/GLUT1DS2)[δ]	SLC2A1[δ]
☐ Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations[δ]	MEF2C[δ]
☐ Nocturnal frontal lobe epilepsy type 1 (ADNFLE1)[δ]	CHRNA4[δ]
☐ Nocturnal frontal lobe epilepsy type 3 (ADNFLE3)[δ]	CHRNB2[δ]
☐ Progressive myoclonic epilepsy type 1A (EPM1) / Unverricht-Lundborg disease (ULD)	CSTB
☐ Progressive myoclonic epilepsy type 1B (EPM1B)	PRICKLE1
☐ Progressive myoclonic epilepsy type 2A (EMP2A)/ Lafora[δ]	EPM2A[δ]
☐ Progressive myoclonic epilepsy type 2B (EPM2B)/ Lafora[δ]	NHLRC1[δ]
☐ Progressive myoclonic epilepsy type 3 (EPM3)	KCTD7
☐ Progressive myoclonic epilepsy type 4, AMRF, (EPM4)	SCARB2
☐ Progressive myoclonic epilepsy type 5 (EPM5)	PRICKLE2
☐ Progressive myoclonic epilepsy type 6 (EPM6)	GOSR2
☐ Pyridoxine-dependent epilepsy (PDE)	ALDH7A1
☐ Pyridoxine-dependent epilepsy (PDE)	PNPO
☐ X-linked Multiple congenital anomalies-hypotonia-seizures syndrome 2	PIGA
☐ X-linked Rolandic epilepsy, mental retardation and speech dyspraxia (RESDX)	SRPX2

Hereditary cancer
Gene panels

☐ **Ovarian cancer** (ONC01v16.1; 2 genes)
BRCA1 and *BRCA2* copy number analysis included
BRCA1, BRCA2, BRIP1, RAD51C, RAD51D

* NGS gene panel analysis can only detect single nucleotide changes and small deletions/duplications. Large copy number changes and repeat expansions cannot be detected. Unless indicated otherwise, these analyses must be requested seperately.

[δ] Sequence and copy number analysis

^ Repeat expansion analysis only

Hereditary cancer (Continued)
Gene panels

☐ **Breast cancer** (ONC02v18.1; 4 genes)
BRCA1 and *BRCA2* copy number analysis included
ATM, BRCA1, BRCA2, CHEK2, PALB2

☐ **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

☐ **MEN1** (ONC06v18.1); 7 genes)
AIP, CDKN1B and MEN1 copy number analysis included.
AIP, CDC73, CDKN1A, CDKN1B, CDKN2B, CDKN2C, MEN1

☐ **Renal cancer** (ONC07v18.1); 7 genes)
VHL copy number analysis included.
BAP1, FH, FLCN, MET, PTEN, SDHB, VHL

☐ **Polyposis/colorectal cancer** (ONC08v20.1; 19 genes)
APC, MUTYH (6 out of 16 exons), promotor region GREM1, BMPR1A, SMAD4 and PTEN copy number analysis included.
APC, BMPR1A, EPCAM, GREM1, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2 (reduced sensitivity due to pseudogene presence)), POLD1, POLE, PTEN, RNF43, RPS20, SMAD4, STK11

☐ **Non-polyposis/colorectal cancer** (ONC09v20.1; 7 genes)
MSH6, MLH1 and MSH. copy number analysis included.
EPCAM, MLH1, MSH2, MSH6, PMS2 (reduced sensitivity due to pseudogene presence), POLD1, POLE

Hereditary cancer
Single gene | Sequence analysis

☐ Acromegaly, Pituitary adenoma predisposition (PAP)[δ]	AIP[δ]
☐ Breast cancer, familial[δ]	BRCA1[δ]
☐ Breast cancer, familial[δ]	BRCA2[δ]
☐ Breast cancer, copy number analysis only	BRCA1
☐ Breast cancer, copy number analysis only	BRCA2
☐ Breast cancer, familial	CHEK2
☐ Breast cancer, familial	PALB2
☐ Oligodontia-colorectal cancer syndrome (ODCRCS)	AXIN2
☐ Emberger syndrome	GATA2
☐ Familial acute myeloid leukemia (AML)[δ]	CEBPA[δ]
☐ Familial acute myeloid leukemia / platelet disorder (AML/FDP)[δ]	RUNX1[δ]
☐ Pheochromocytoma / paraganglioma (FEO/PGL)[δ]	SDHB[δ]
☐ Pheochromocytoma / paraganglioma (FEO/PGL)[δ]	SDHC[δ]
☐ Pheochromocytoma / paraganglioma (FEO/PGL)[δ]	SDHD[δ]
☐ Pheochromocytoma / paraganglioma (FEO/PGL)	TMEM127
☐ Pheochromocytoma / paraganglioma (FEO/PGL)	MAX
☐ Hyperparathyroidism, familiar primary (HRPT1)[δ]	MEN1[δ]
☐ 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 2A (MEN2A) (*MEN2A relevant exons only)*	RET
☐ Multiple endocrine neoplasia type 4[δ]	CDKN1B[δ]
☐ Multiple endocrine neoplasia, atypical	CDKN1A
☐ Multiple endocrine neoplasia, atypical	CDKN2B
☐ Multiple endocrine neoplasia, atypical	CDKN2C
☐ Papillary renal cell carcinoma, familial (HPRC)	MET
☐ Sporadic medullary thyroid carcinoma (SMTC)	RET
☐ Von Hippel-Lindau disease (VHL)[δ]	VHL[δ]

Intellectual disability: syndromal/non-syndromal
Gene panel | Exome

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

Intellectual disability | gene panel/exome (VBE01v18.1; 989 genes/exome)
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

☐ Albright hereditary osteodystrophy (AHO) *(sequence-analysis and methylation specific copy number analysis)*	GNAS
☐ Angelman syndrome (AS) *(methylation specific copy number analysis)*	[15q11-q13]
☐ Angelman syndrome (AS)[δ]	UBE3A[δ]
☐ Cohen syndrome[δ] *[OBE01v16.1]*	VPS13B[δ]
☐ Fragile-X syndrome (FRAX), FRAXA included^	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 specific copy number analysis)*	[15q11-q13]
☐ Pseudohypoparathyroidism, type 1A (PHP1A)[δ] *(sequence-analysis and methylation specific copy number analysis)*	GNAS
☐ X-linked intellectual disability	HDAC8

Metabolic diseases
Gene panels

☐ **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

☐ **Intrahepatic cholestasis** (MET02v16.2; 5 genes)
ATP8B1, ABCB11, ABCB4, TJP2, NR1H4

☐ **Mitochondrial resperatory chain diseases** (MET07v16.1; 32 genes)
ADCK3, ANTI, APTX, BCS1L, C10ORF2, C12ORF62, C2ORF64, COQ2, COQ9, COX6B1, DGUOK, FASTKD2, NDUFAF2, NDUFAF3, NDUFAF4, NDUFB3, NDUFS1, NDUFS2, NDUFS4, NDUFS6, OPA1, PDSS1, PDSS2, POLG, RRM2B, SDHA, SDHAF1, SUCLA2, TK2, TTC19, UQCRB, UQCRQ

☐ **Serine synthesis defect** (MET03v16.1; 3 genes)
PHGDH, PSPH, PSAT1

☐ **Fatty acid oxidation disease** (MET05v15.1; 12 genes)
ACADVL, CPT1A, CPT1B, CPT2, ETFA, ETFB, ETFHD, HADHA, HADHB, SLC22A5, SLC25A20, SLC52A3

* NGS gene panel analysis can only detect single nucleotide changes and small deletions/duplications. Large copy number changes and repeat expansions cannot be detected. Unless indicated otherwise, these analyses must be requested seperately.

[δ] Sequence and copy number analysis

^ Repeat expansion analysis only

Metabolic diseases (Continued)
Single gene | Sequence analysis

☐ **Neonatal and paediatric cholestasis** (MET09v16.2; 26 genes)
ABCB11, ABCB4, ABCC2, ATP7B, ATP8B1, BCS1L, C10ORF2, CFTR, CIRH1A, DGUOK, FAH, GALT, JAG1, MPV17, NOTCH2, NPC1, NPC2, POLG, SCO1, SERPINA1, SLC25A13, SUCLA2, TALDO1, TJP2, NR1H4, CYP27A1

☐ **Niemann-Pick disease** (MET04v16.1; 3 genes)
SMPD1, NPC1, NPC2

☐ **Syndromes with cholestasis** (MET10v16.2; 63 genes)
ABCB11, ABCB4, ABCC2, ABCD3, ADK, AHCY, AKR1D1, ALDOB, AMACR, ARG1, ASAH1, ATP7B, ATP8B1, BAAT, BCS1L, C10ORF2, CFTR, CIRH1A, CLDN1, CYP7B1, DCDC2, DGUOK, DHCR7, FAH, GALT, GBA, GBE1, GLIS3, HADHA, HNF1A, HNF1B, HSD3B7, IFT43, INVS, JAG1, LIPA, MPV17, MTM1, MYO5B, NOTCH2, NPC1, NPC2, NPHP3, PEX1, PEX14, POLG, POMC, PROP1, SCO1, SERPINA1, SHPK, SLC25A13, SLC27A5, STX3, SUCLA2, TALDO1, TJP2, TPO, TRMU, VIPAS39, VPS33B, NR1H4, CYP27A1

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
☐ Hyperinsulinemic hypoglycemia, familial, type 7 (HHF7)	SLC16A1
☐ Phenylketonuria type 1 (PKU)	PAH
☐ Phenylketonuria type 3 (PTPS)	PTS
☐ Glycerol kinase deficiency (GKD)[δ]	GK[δ]
☐ Glycine encephalopathy / nonketotic hyperglycinemia	AMT
☐ Glycine encephalopathy / nonketotic hyperglycinemia	GCSH
☐ Glycine encephalopathy / nonketotic hyperglycinemia[δ]	GLDC[δ]
☐ Hartnup disorder	SLC6A19
☐ Hemochromatosis, (HFE)	HFE
☐ Intrahepatic cholestasis type 1, BRIC/PFIC type 1	ATP8B1
☐ Intrahepatic cholestasis type 2, BRIC/PFIC type 2	ABCB11
☐ Intrahepatic cholestasis type 3, BRIC/PFIC type 3	ABCB4
☐ Medium-Chain Acyl-CoA dehydrogenasedeficiency	ACADM
☐ Metachromatic leukodystrophy (MLD)[δ]	ARSA[δ]
☐ Methylmalonic aciduria type cblA	MMAA
☐ Pompe disease, Glycogen storage disease II (GSD2)	GAA
☐ Pyruvate kinase deficiency (PK)	PKLR
☐ Serine biosynthesis defect, PHGDH deficiency	PHGDH
☐ Serine biosynthesis defect, PSPH deficiency	PSPH
☐ Serine biosynthesis defect, PSAT1 deficiency	PSAT1
☐ Tyrosinemia, type I	FAH
☐ Wilson disease (WD)[δ]	ATP7B[δ]

Neurological disorders
Gene panels

See Neuromuscular diseases for the Ataxia NGS panel

☐ **FTD-ALS*** (NEU01v17.1; 16 genes)
ALS2, ANG, CHMP2B, FIG4, FUS, GRN, MAPT, NPC1, NPC2, SETX, SMPD1, SOD1, TARDBP, UB1LN2, VAPB, VCP
Repeat expansion analysis*: ☐ C9ORF72

☐ **Cerebral cavernous malformations (CCM)** (NEU03v16.1; 3 genes)
Includes copy number analysis of KRIT1, CCM2 and PDCD10
KRIT1, CCM2, PDCD10

Neurological disorders
Single gene | Sequence / repeat expansion analysis

☐ Amyloidosis I and VII; transthyretin amyloidosis	TTR
☐ Amyotrophic lateral sclerosis type 1 (ALS1)	SOD1
☐ Amyotrophic lateral sclerosis (Juvenile) type 2 (ALS2)	ALS2
☐ Amyotrophic lateral sclerosis type 4 (ALS4)	SETX
☐ Amyotrophic lateral sclerosis type 6 (ALS6)	FUS
☐ Amyotrophic lateral sclerosis type 8 (ALS8)	VAPB
☐ Amyotrophic lateral sclerosis type 9 (ALS9)	ANG
☐ Amyotrophic lateral sclerosis type 10 (ALS10)	TARDBP
☐ Amyotrophic lateral sclerosis type 11 (ALS11)	FIG4
☐ Amyotrophic lateral sclerosis type 14 (ALS14)	VCP
☐ Amyotrophic lateral sclerosis type 15 (ALS15), with or without FTD	UBQLN2
☐ Amyotrophic lateral sclerosis/ Frontotemporal dementia (FTDALS)^	C9ORF72^
☐ Cerebral cavernous malformations type 1 (CCM1)[δ]	KRIT1[δ]
☐ Cerebral cavernous malformations type 2 (CCM2)[δ]	CCM2[δ]
☐ Cerebral cavernous malformations type 3 (CCM3)[δ]	PDCD10[δ]
☐ Frontotemporal dementia (FTD)[δ]	MAPT[δ]
☐ Frontotemporal dementia (FTD)[δ]	GRN[δ]
☐ Fuhrmann syndrome	WNT7A
☐ Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia	VCP
☐ Pitt Hopkins-like syndrome 1	CNTNAP2
☐ Pitt Hopkins-like syndrome 2[δ]	NRXN1[δ]
☐ Schizencephaly(CBPS)	EMX2
☐ Spinocerebellar ataxia type 1 (SCA1)^	ATXN1^
☐ Spinocerebellar ataxia type 2 (SCA2)^	ATXN2^
☐ Spinocerebellar ataxia type 3 (SCA3)^	ATXN3^
☐ Spinocerebellar ataxia type 6 (SCA6)^	CACNA1A^
☐ Spinocerebellar ataxia type 7 (SCA7)^	ATXN7^
☐ Spinocerebellar ataxia type 12 (SCA12)^	PPP2R2B^
☐ Spinocerebellar ataxia type 13 (SCA13)	KCNC3
☐ Spinocerebellar ataxia type 14 (SCA14)	PRKCG
☐ Spinocerebellar ataxia type 17 (SCA17)^	TBP^
☐ Spinocerebellar ataxia type 23 (SCA23)	PDYN
☐ Spinocerebellar ataxia type 28 (SCA28)	AFG3L2

Neuromuscular disease
Gene panels

• Repeat expansions and (larger) copy number changes 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.

☐ **Ataxia•** (NEM14v19.1; 43 genes)
ADCK3, AFG3L2, APTX, ATM, BEAN1, CACNA1A, CACNA1G, CACNB4, CCDC88C, EEF2, ELOVL4, ELOVL5, FGF14, FXN, IFRD1, ITPR1, KCNA1, KCNC3, KCND3, MME, MRE11A, NOP56, PDYN, PEX7, PHYH, POLG, PRKCG, RNF216, SACS, SETX, SIL1, SLC1A3, SPTBN2, STUB1, SYNE1, TDP1, TGM6, TK2, TMEM240, TRPC3, TTBK2, TTPA, TWNK
Repeat expansion analysis•: ☐ ATXN1 ☐ ATXN2 ☐ ATXN3 ☐ ATXN7 ☐ CACNA1A ☐ PPP2R2B ☐ TBP ☐ FMR1 (FXTAS)

☐ **Congenital/metabolic myasthene syndromes** (NEM12v19.1; 31 genes)
AGRN, ALG14, ALG2, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COL13A1, COLQ, DOK7, DPAGT1, GFPT1, GMPPB, LAMA5, LAMB2, LRP4, MUSK, MYO9A, PLEC, PREPL, RAPSN, SCN4A, SLC18A3, SLC25A1, SLC5A7, SNAP25, SYT2, TPM3, VAMP1

* NGS gene panel analysis can only detect single nucleotide changes and small deletions/duplications. Large copy number changes and repeat expansions cannot be detected. Unless indicated otherwise, these analyses must be requested seperately.

[δ] Sequence and copy number analysis

^ Repeat expansion analysis only

Neuromuscular diseases (Continued)
Gene panels

☐ **Congenital muscular dystrophy** (NEM07v19.1; 34 genes)
ACTA1, ALG13, B3GALNT2, B3GNT1, CHKB, COL12A1, COL6A1, COL6A2, COL6A3, DAG1, DNM2, DPM1, DPM2, FHL1, FKRP, FKTN, GMPPB, GOLGA2, INPP5K, ISPD, ITGA7, LAMA2, LARGE, LMNA, POMGNT1, POMGNT2, POMK, POMT1, POMT2, SELENON, TCAP, TMEM5, TRAPPC11, TRIP4

☐ **Congenital myopathy** (NEM04v19.1; 32 genes)
ACTA1, BIN1, CACNA1S, CFL2, CNTN1, DNM2, HNRNPA1, HRAS, KBTBD13, KLHL40, KLHL41, LMOD3, MAP3K20, MEGF10, MTM1, MYBPC3, MYH2, MYH7, MYMK, MYO18B, MYPN, NEB, PTPLA, RYR1, SELENON, SPEG, SPTBN4, TNNT1, TPM2, TPM3, TRIM32, TTN

☐ **Distal myopathy** (NEM05v19.1; 21 genes)
ADSSL1, ANO5, BAG3, CAV3, CRYAB, DES, DNM2, DYSF, FLNC, GNE, KLHL9, KY, LDB3, MATR3, MYH7, MYOT, NEB, SELENON, TIA1, TTN, VCP

☐ **Hereditary spastic paraplegia (HSP)** (NEM26v19.1; 57 genes)
ATL1 and SPAST copy number analysis included
AFG3L2, ALDH18A1, ALDH3A2, ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATL1, B4GALNT1, BSCL2, C12orf65, C19orf12, CAPN1, CYP2U1, CYP7B1, DDHD1, DDHD2, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, GBA2, GJC2, HSPD1, IBA57, KIAA0196, KIF1A, KIF1C, KIF5A, L1CAM, MAG, MARS2, MTPAP, NIPA1, NT5C2, PLP1, PNPLA6, REEP1, RTN2, SACS, SLC33A1, SPAST, SPG11, SPG20, SPG21, SPG7, TECPR2, TFG, VAMP1, VPS37A, ZFYVE26, ZFYVE27

☐ **Limb-Girdle muscle weakness** (NEM08v19.2; 42 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, PTRF, 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) (NEM13v19.1; 55 genes)
AARS, ALS2, ANG, AR, ASAH1, ASCC1, ATP7A, BICD2, BSCL2, CHCHD10, CHMP2B, DCTN1, DNAJB2, DYNC1H1, ERBB3, ERBB4, EXOSC3, EXOSC8, FBXO38, FIG4, FUS, GARS, GLE1, HEXB, HNRNPA1, HSPB1, HSPB3, IGHMBP2, MATR3, NEFH, OPTN, PFN1, PIP5K1C, PLEKHG5, PRPH, RBM7, REEP1, SETX, SIGMAR1, SLC52A2, SLC52A3, SLC5A7, SOD1, SPG11, SQSTM1, TARDBP, TRIP4, TRPV4, TUBA4A, UBA1, UBQLN2, VAPB, VCP, VRK1, WARS
Repeat expansion analysis●: ☐ C9ORF72
Copy number analysis●: ☐ SMN1(/SMN2)

☐ **Motor and Sensory Neuropathy**● (NEM15v19.1; 88 genes)
AARS, AIFM1, ARHGEF10, ATL1, ATL3, BAG3, BSCL2, CCT5, COX6A1, CTDP1, DCAF8, DGAT2, DHTKD1, DNAJB2, DNM2, DNMT1, DST, DYNC1H1, EGR2, FAM134B, FBLN5, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, GJB3, GNB4, HARS, HINT1, HK1, HOXD10, HSPB1, HSPB3, HSPB8, IGHMBP2, IKBKAP, INF2, KARS, KIF1A, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, MARS, MED25, MFN2, MME, MORC2, MPZ, MTMR2, NAGLU, NDRG1, NEFH, NEFL, NGF, NTRK1, PDK3, PLEKHG5, PMP2, PMP22, PNKP, PRDM12, PRPS1, PRX, RAB7A, SBF1, SBF2, SCN11A, SCN9A, SGPL1, SEPT9, SH3TC2, SLC12A6, SPG11, SPTLC1, SPTLC2, SURF1, TFG, TRIM2, TRPV4, TTR, VCP, VRK1, WNK1, YARS
Copy number analysis●: ☐ PMP22/MPZ/GJB1

☐ **Myotonic syndromes**● (NEM09v16.1; 7 genes)
ATP2A1, CAV3, CLCN1, CNBP, DMPK, HSPG2, SCN4A
Repeat expansion analysis●: ☐ DMPK ☐ CNBP

☐ **NMDs affecting the peripheral nervous system** (NEM27v19.2; 290 genes)
AARS, ACTA1, ACVR1, ADSSL1, AGRN, AIFM1, ALG13, ALG14, ALG2, ALS2, ANG, ANO5, AR, ARHGEF10, ASAH1, ASCC1, ATL1, ATL3, ATP2A1, ATP7A, B3GALNT2, B3GNT1, BAG3, BICD2, BIN1, BSCL2, BVES, CACNA1S, CAPN3, CASQ1, CAV3, CCT5, CFL2, CHAT, CHCHD10, CHKB, CHMP2B, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CLCN1, CLN3, CNBP, CNTN1, CNTNAP1, COL12A1, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, COX6A1, CRYAB, CTDP1, DAG1, DCAF8, DCTN1, DES, DGAT2, DHTKD1, DMD, DMPK, DNAJB2, DNAJB6, DNM2, DNMT1, DOK7, DPAGT1, DPM1, DPM2, DPM3, DST, DYNC1H1, DYSF, EGR2, EMD, ERBB3, ERBB4, EXOSC3, EXOSC8, FAM111B, FAM134B, FASTKD2, FBLN5, FBXO38, FGD4, FHL1, FIG4, FKRP, FKTN, FLNC, FUS, GAA, GAN, GARS, GDAP1, GFPT1, GJB1, GJB3, GLE1, GMPPB, GNB4, GNE, GOLGA2, HARS, HEXB, HINT1, HK1, HNRNPA1, HNRNPDL, HOXD10, HRAS, HSPB1, HSPB3, HSPB8, HSPG2, IGHMBP2, IKBKAP, INF2, INPP5K, ISCU, ISPD, ITGA7, KARS, KBTBD13, KIF1A, KIF1B, KIF21A, KIF5A, KLHL40, KLHL41, KLHL9, KY, LAMA2, LAMA5, LAMB2, LARGE, LDB3, LIMS2, LITAF, LMNA, LMOD3, LRP4, LRSAM1, MAP3K20, MARS, MATR3, MED25, MEGF10, MFN2, MME, MORC2, MPZ, MSTN, MTM1, MTMR2, MUSK, MYBPC3, MYH2, MYH3, MYH7, MYH8, MYMK, MYO18B, MYO9A, MYOT, MYPN, NAGLU, NDRG1, NEB, NEFH, NEFL, NGF, NTRK1, OPA1, OPTN, ORAI1, PABPN1, PDK3, PFN1, PHOX2A, PIP5K1C, PLEC, PLEKHG5, PMP2, PMP22, PNKP, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PRDM12, PREPL, PRPH, PRPS1, PRX, PTPLA, PTRF, PTRH2, PUS1, PYGM, PYROXD1, RAB7A, RAPSN, RBM7, REEP1, RRM2B, RYR1, SBF1, SBF2, SCN11A, SCN4A, SCN9A, SELENON, SEPT9, SETX, SGCA, SGCB, SGCD, SGCE, SGCG, SGPL1, SH3TC2, SIGMAR1, SLC12A6, SLC25A4, SLC25A42, SLC52A2, SLC52A3, SLC5A7, SMCHD1, SNAP25, SOD1, SPEG, SPG11, SPTBN4, SPTLC1, SPTLC2, SQSTM1, STIM1, SUCLA2, SURF1, SYNE1, SYNE2, SYT2, TARDBP, TCAP, TFG, TIA1, TK2, TMEM43, TMEM5, TMEM65, TNNI2, TNNT1, TNNT3, TNPO3, TOR1A, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM2, TRIM32, TRIM54, TRIM63, TRIP4, TRPV4, TTN, TTR, TUBA4A, TUBB3, TWNK, UBA1, UBQLN2, VAMP1, VAPB, VCP, VMA21, VRK1, WARS, WNK1, YARS, YARS2

☐ **NMDs with episodic attacks** (NEM28v19.1; 14 genes)
CACNA1A, CACNA1S, CLCN1, KCNA1, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ18, KCNJ2, KCNQ1, RYR1, SCN4A, SCN5A

☐ **Periodic paralysis and ion channel muscle disease** (NEM10v19.1; 13 genes)
CACNA1A, CACNA1S, CLCN1, KCNA1, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ18, KCNJ2, KCNQ1, SCN4A, SCN5A

☐ **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, FAM111B, FASTKD2, IKBKAP, 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/malignant hyperthermia *[NEM29v19.1]*	RYR1
☐ Ehlers-Danlos syndrome (musculocontractural)	CHST14
☐ Kennedy Disease; SBMA, X-linked Type 1 (SMAX1)^	AR^
☐ Motor and sensory neuropathy *(copy number analysis only)*	PMP22/MPZ/GJB1
☐ Muscular dystrophy, Emery-Dreifuss type 6 (EDMD6)	FHL1
☐ Muscular dystrophy, Limb-Girdle type 2G (LGMD2G)	TCAP
☐ Myofibrillar myopathy type 1 (MFM1)	DES
☐ Myofibrillar myopathy type 2 (MFM2)	CRYAB
☐ Myotonic dystrophy type 1 (DM1)^	DMPK^
☐ Myotonic dystrophy type 2 (DM2)^	CNBP^
☐ Nemaline myopathy type 1 (NEM1)	TPM3
☐ Nemaline myopathy type 3 (NEM3)	ACTA1
☐ Nemaline myopathy type 4 (NEM4)	TPM2
☐ Nemaline myopathy type 5 (NEM5)	TNNT1
☐ Nemaline myopathy type 6 (NEM6)	KBTBD13
☐ Nemaline myopathy type 7 (NEM7)	CFL2
☐ Spinal Muscular Atrophy (SMA type 1 - 4)$^{\delta}$ *(sequence analysis only after consultation)*	SMN1$^{\delta}$

* NGS gene panel analysis can only detect single nucleotide changes and small deletions/duplications. Large copy number changes and repeat expansions cannot be detected. Unless indicated otherwise, these analyses must be requested seperately.

$^{\delta}$ Sequence and copy number analysis

^ Repeat expansion analysis only

Obesity
Single gene | Sequence analysis

☐ Cohen syndrome[δ]	*[OBE01v16.1]*	VPS13B[δ]
☐ Leptin deficiency		LEP
☐ Leptin receptor deficiency		LEPR
☐ Obesity with impaired prohormone processing		PCSK1
☐ Proopiomelanocortin deficiency		POMC
☐ Obesity, autosomal dominant		MC4R

Primary immunodeficiencies
Gene panels

☐ **Autoinflammatory disease*** (PID01v17.2; 33 genes)
AP1S3, CARD14, CECR1, IL10, IL10RA, IL10RB, IL1RN, IL36RN, LPIN2, MEFV, MVK, NCSTN, NLRC4, NLRP1, NLRP12, NLRP3, NLRP7, NOD2, OTULIN, PLCG2, PSENEN, PSMA3, PSMB4, PSMB8, PSMB9, PSTPIP1, RBCK1, SH3BP2, SLC29A3, TMEM173, TNFAIP3, TNFRSF11A, TNFRSF1A
Copy number analysis*: ☐ IL1RN ☐ IL10RB

☐ **HLH/Immune dysregulation*** (PID02v16.1; 9 genes)
PRF1, UNC13D, STX11, STXBP, SH2D1A, XIAP, LYST, RAB27A, AP3B
Copy number analysis*: ☐ PRF1 ☐ UNC13D ☐ STX11

☐ **ALPS/Autoimmunity** (PID03v17.1; 12 genes)
FAS, FASLG, CASP10, CASP8, KRAS, NRAS, FADD, AIRE, FOXP3, IL2RA, ITCH, LRBA

☐ **(S)CID** (PID04v16.1**;** 27 genes)
Includes copy number analysis of DOCK8
ADA, AK2, CD3D, CD3E, CD3G, CD40, CD8A, CORO1A, DCLRE1C, IL2RA, Il2RG, IL7R, JAK3 LIG4, NHEJ1, PNP, PRKDC, PTPRC, RAG1, RAG2, ZAP70, CD40LG, ORAI1, STIM1, STAT5B, DOCK8, TBX1

☐ **B-cell pathology** (PID05v16.1; 14 genes)
BTK, ICOS, CD19, CD81, TNFRSF13B, TNFRSF13C, CD40, CD40L, AICDA, UNG, CD79A, BLNK, CD79B, IGLL1

☐ **HIES syndromes** (PID06v16.1; 3 genes)
Includes copy number analysis of DOCK8
STAT3, TYK2, DOCK8

☐ **Chronic mucocutaneous candidiasis (CMC)** (PID07v17.1; 7 genes)
Il17RA, IL17F, STAT1, TLR3, AIRE, IL2RA, CARD9

☐ **Primary immunodeficiencies full panel** (PID00v20.1; 420 genes)
ACD, ACP5, ACTB, ADA, ADA2, ADAM17, ADAR, AGA, AICDA, AIRE, AK2, ALG13, ALPI, AP1S3, AP3B1, AP3D1, APOL1, ARHGEF1, ARPC1B, ATM, ATP6AP1, B2M, BACH2, BCL10, BCL11B, BLK, BLM, BLNK, BLOC1S6, BTK, C17orf62, C1QA, C1QB, C1QC, C1R, C1S, C2, C3, C5, C6, C7, C8A, C8B, C8G, C9, CA2, CARD11, CARD14, CARD9, CARMIL2, CASP10, CASP8, CAVIN1, CCBE1, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD46, CD55, CD59, CD70, CD79A, CD79B, CD81, CD8A, CDCA7, CDKN2B, CEBPE, CFB, CFD, CFH, CFHR1, CFHR3, CFHR5, CFI, CFP, CFTR, CHD7, CIB1, CIITA, CLCN7, CLEC4D, CLEC7A, CLPB, COPA, CORO1A, CR2, CREBBP, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTPS1, CTSC, CXCR4, CYBA, CYBB, DBR1, DCLRE1B, DCLRE1C, DDX58, DEF6, DGAT1, DHFR, DKC1, DNAJC21, DNASE1, DNASE1L3, DNASE2, DNMT3B, DOCK2, DOCK8, ELANE, ELF4, EPG5, ERCC2, ERCC3, ERCC6L2, EXTL3, F12, FAAP24, FADD, FAS, FASLG, FAT4, FCGR1A, FCGR2A, FCGR2B, FCGR3A, FCGR3B, FCHO1, FCN3, FERMT3, FOXN1, FOXP3, FPR1, G6PC, G6PC3, G6PD, GATA2, GFI1, GINS1, GJC2, GRHL2, GTF2H5, HAVCR2, HAX1, HELLS, HMOX1, HYOU1, ICOS, ICOSLG, IFIH1, IFNAR1, IFNAR2, IFNGR1, IFNGR2, IGHM, IGLL1, IKBKB, IKBKG, IKZF1, 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, JAGN1, JAK1, JAK2, JAK3, KDM6A, KMT2D, LACC1, LAMTOR2, LAT, LCK, LIG1, LIG4, LPIN2, LRBA, LRRC8A, LTBP3, LYST, MAGT1, MAL, MALT1, MAN2B1, MANBA, MAP3K14, MASP2, MBL2, MC2R, MCM4, MEFV, MKL1, MOGS, MRE11, MS4A1, MSN, MTHFD1, MVK, MYD88, MYSM1, NBAS, NBN, NCF1, NCF2, NCF4, NCSTN, NFAT5, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, NOP10, NRAS, NSMCE3, OAS1, ORAI1, OSTM1, OTULIN, PARN, PAX5, PBX1, PCCA, PCCB, PEPD, PGM3, PIGA, PIK3CD, PIK3R1, PLCG2, PLEKHM1, PLG, PMM2, PNP, POLA1, POLE2, POMP, POT1, PRF1, PRKCD, PRKDC, PRPS1, PSENEN, PSMA3, PSMB4, PSMB8, PSMB9, PSMG2, PSTPIP1, PTPN22, PTPRC, RAB27A, RAC2, RAG1, RAG2, RANBP2, RASGRP1, RASGRP2, RBCK1, RECQL4, RELB, RFX5, RFXANK, RFXAP, RHOH, RIPK1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RNU4ATAC, RORC, RPSA, RSPH9, RTEL1, SAMD9, SAMD9L, SAMHD1, SBDS, SEC61A1, SEMA3E, SERAC1, SERPING1, SH2B3, SH2D1A, SH3BP2, SH3KBP1, SKIV2L, SLC29A3, SLC35A1, SLC35C1, SLC37A4, SLC39A4, SLC39A7, SLC46A1, SLC7A7, SMARCAL1, SMARCD2, SNX10, SOCS4, SP110, SPINK5, SPPL2A, SRP54, SRP72, STAT1, STAT2, STAT3, STAT4, STAT5B, STAT6, STIM1, STK4, STN1, STX11, STXBP2, TAP1, TAP2, TAPBP, TAZ, TBX1, TCF3, TCIRG1, TCN2, TERC, TERT, TFRC, TGFB1, THBD, TICAM1, TINF2, TIRAP, TLR3, TLR4, TMC6, TMC8, TMEM173, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF4, TNFRSF9, TNFSF11, TNFSF12, TOP2B, TPP2, TRAC, TRAF3, TRAF3IP2, TREX1, TRIM22, TRNT1, TTC37, TTC7A, TYK2, UNC13D, UNC93B1, UNG, USB1, USP18, VAV1, VPS13B, VPS45, WAS, WDR1, WIPF1, WRAP53, XIAP, ZAP70, ZBTB24, ZNF341

Primary immunodeficiencies
Single gene | Sequence analysis

☐ Acne inversa, familiar type 1	NCSTN
☐ Acne inversa, familiar type 2	PSENEN
☐ ADA2 deficieny	CECR1
☐ Agammaglobulinemia, X-linked (XLA)	BTK
☐ Autoimmune lymphoproliferative syndrome, (ALPS), type 1a[δ]	FAS[δ]
☐ Autoimmune lymphoproliferative syndrome, (ALPS), type 1b	FASL
☐ Autoimmune lymphoproliferative syndrome, (ALPS), type 2a	CASP10
☐ Autoimmune polyendocrinopathy syndrome, type I (APS1)	AIRE
☐ Blau syndrome	NOD2
☐ CINCA syndrome	NLRP3
☐ Candidiasis, familiar type 2	CARD9
☐ Candidiasis, familiar type 5	IL17RA
☐ Candidiasis, familiar type 6	IL17F
☐ Candidiasis, familiar type 7	STAT1
☐ Cold-induced autoinflammatory syndrome (FCAS1)	NLRP3
☐ Cold-induced autoinflammatory syndrome (FCAS2)	NLRP12
☐ Cold-induced autoinflammatory syndrome (FCAS3)[δ]	PLCG2[δ]
☐ DIRA syndrome[δ]	IL1RN[δ]
☐ Familial Mediterranean fever (FMF)	MEFV
☐ Hydatidiform mole, recurrent type 1	NLRP7
☐ Hemophagocytic lymphohistiocytosis, HLH type 2[δ]	PRF1[δ]
☐ Hemophagocytic lymphohistiocytosis, HLH type 3[δ]	UNC13D[δ]
☐ Hemophagocytic lymphohistiocytosis, HLH type 4[δ]	STX11[δ]
☐ Hemophagocytic lymphohistiocytosis, HLH type 5	STXBP2
☐ Hyper-IgM syndrome, CD40 ligand deficiency	CD40LG
☐ Hyper-IgM syndrome, AID deficiency	AICDA
☐ Hereditary Angiodema type 1	SERPING1
☐ Hyper-IgE syndrome[δ]	DOCK8[δ]
☐ Hyper-IgE syndrome[δ]	STAT3[δ]
☐ Hyper-IgD syndrome (HIDS)	MVK
☐ Inflammatory Bowel Disease (IBD)	IL10RA
☐ Inflammatory Bowel Disease (IBD)[δ]	IL10RB[δ]
☐ JPM syndrome, Candle syndrome, Nakajo syndrome	PSMB8
☐ Mevalonate kinase deficiency (MKD)	MVK
☐ Muckle-Wells syndrome	NLRP3
☐ Multiple congenital anomalies-hypotonia-seizures syndrome 2	PIGA
☐ PAPA syndrome	PSTPIP1
☐ Psoriasis, generalized pustular[δ]	IL36RN[δ]
☐ Severe combined immunodeficiency (SCID), X-linked, Common γ chain deficiëntie	IL2RG
☐ Severe combined immunodeficiency (SCID)	ZAP70
☐ Severe combined immunodeficiency (SCID)	CD3G
☐ Severe combined immunodeficiency (SCID)	CD3D

* NGS gene panel analysis can only detect single nucleotide changes and small deletions/duplications. Large copy number changes and repeat expansions cannot be detected. Unless indicated otherwise, these analyses must be requested seperately.

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

Primary immunodeficiencies (Continued)
Single gene | Sequence analysis

☐ Severe combined immunodeficiency (SCID)	CD3E
☐ Severe combined immunodeficiency (SCID)	RAG1
☐ Severe combined immunodeficiency (SCID)	RAG2
☐ TNFR associated periodic fever syndrome (TRAPS)	TNFRSF1A
☐ WHIM syndrome	CXCR4
☐ Wiskott-Aldrich syndrome	WAS
☐ X-linked lymphoproliferative syndrome, type 1 (XLP1)[δ]	SH2D1A[δ]
☐ X-linked lymphoproliferative syndrome, type2 (XLP2)	XIAP

Renal disease
Gene panels

See Hereditary cancer for the renal cancer panel.

☐ **Atypical Hemolytic uremic syndrome (aHUS)/ Thrombotic microangiopathies** (NEF07v18.1; 12 genes)
Includes copy number analysis of CD46, CFH, CFI
ADAMTS13, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFI, DGKE, THBD

☐ **Alport syndrome** (NEF01v.16.1; 3 genes)
COL4A3, COL4A4, COL4A5

☐ **Alport syndrome, broad differential diagnosis** (NEF23v18.1; 19 genes)
ACTN4, C3, CD2AP, CFH, CFHR5, COL4A1, COL4A3, COL4A4, COL4A5, FN1, INF2, LMX1B, MYH9, MYO1E, NPHS1, NPHS2, SLC7A7, TRPC6, WT1

☐ **Congenital anomalies of the kidney and urinary tract (CAKUT)*** (NEF03v18.1; 63 genes)
ACE, ACTG2, AGT, AGTR1, ANO1, BICC1, BMP4, CHD1L, CHD7, CHRM3, DSTYK, EYA1, FAM58A, FGF20, FGF8, FOXF1, FRAS1, FREM1, FREM2, GATA3, GDNF, GLI3, GREB1L, GRIP1, HAAO, HNF1B, HOXD13, HPSE2, ITGA8, JAG1, KAL1, KIF14, KYNU, LMOD1, LPP, LRIG2, LRP4, MKKS, MYH11, NOTCH2, NPHP1, NPHP3, NPHP4, PAX2, PAX8, REN, RET, ROBO2, SALL1, SALL4, SIX1, SIX2, SIX5, SLIT2, SOX17, STRA6, TBC1D1, TRAP1, UMOD, WNT4, WT1, ZEB2, ZIC3
Copy number analysis*: ☐ EYA1 ☐ HNF1B ☐ NPHP1 ☐ PAX2 ☐ RET

☐ **Renal cysts and/or ciliopathies, incl. Bardet-Biedl syndrome,** Nephronophthisis **and Joubert syndrome*** (NEF17v18.1; 115 genes)
Includes copy number analysis of NPHP1
AGXT, AHI1, ALG8, ANKS3, ANKS6, ARL13B, ARL6, ATXN10, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BICC1, C2CD3, C5orf42, CC2D2A, CCDC114, CDKN1C, CEP120, CEP164, CEP290, CEP41, CEP83, COL4A1, CPT2, CRB2, CSPP1, DCDC2, DDX59, DNAJB11, DYNC2H1, DYNC2LI1, DZIP1L, EVC, EVC2, FAN1, GANAB, GLIS2, GLIS3, GPC3, HNF1B, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT57, IFT80, IFT81, INPP5E, INVS, IQCB1, KIAA0556, KIAA0586, KIF14, KIF7, LZTFL1, MAP7D3, MAPKBP1, MKKS, MKS1, MUC1, NEK1, NEK8, NPHP1, NPHP3, NPHP4, OFD1, PBX1, PDE6D, PKD1, PKD2, PKHD1, PMM2, RMND1, RPGRIP1, RPGRIP1L, SCLT1, SDCCAG8, SEC61A1, SEC61B, SLC41A1, SLC4A1, TBX18, TCTEX1D2, TCTN1, TCTN2, TCTN3, TMEM104, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TRAF3IP1, TRIM32, TTC21B, TTC8, UMOD, WDPCP, WDR19, WDR34, WDR35, WDR60, XPNPEP3, ZNF423
Copy number analysis*: ☐ HNF1B

☐ **Renal cysts in adulthood / autosomal dominant tubulointerstitual kidney disaese (ADTKD)** (NEF26v18.1; 20 genes)
ALG8, COL4A1, DNAJB11, GANAB, HNF1B, MUC1, OFD1, PKD1, PKD2, PKHD1, PRKCSH, REN, SEC61A1, SEC61B, SEC63, TMEM104, TSC1, TSC2, UMOD, VHL

☐ **Nephrotic syndrome (NPHS) / Focal segmental glomerulosclerosis (FSGS)** (NEF11v18.1; 74 genes)
ACTN4, ADCK3, ADCK4, ALG1, ANLN, APOL1, ARHGAP24, ARHGDIA, CD151, CD2AP, CFH, CLCN5, COL4A3, COL4A4, COL4A5, COQ2, COQ4, COQ6, COQ7, COQ9, CRB2, CUBN, DGKE, EMP2, FAT1, FN1, FOXC2, GLA, GPC5, GSN, INF2, ITGA3, ITGB4, KANK1, KANK2, KANK4, LAGE3, LAMB2, LCAT, LMNA, LMX1B, LYZ, MAFB, MAGI2, MYH9, MYO1E, NPHS1, NPHS2, NUP107, NUP205, NUP93, NXF5, OSGEP, PAX2, PDSS1, PDSS2, PLCE1, PMM2, PODXL, PTPRO, SCARB2, SEC61A1, SLC7A7, SMARCAL1, SMARCAL1, TP53RK, TPRKB, TRPC6, TTC21B, WDR73, WT1, XPO5, YRDC, ZMPSTE24

☐ **Chronic kidney disease of the young (CKD-Y) (includes PKD1 and PKD2)** (NEF24v18.1; 141 genes)
ACE, ACTN4, ADCK4, AGT, AGTR1, AGXT, ALG1, AMN, ANKS6, APOA1, APOL1, ARHGDIA, ATXN10, B2M, BBIP1, BCS1L, C3, CD151, CD2AP, CD46, CEP164, CEP290, CFB, CFH, CFHR5, CFI, CHD7, CLCN5, COL4A3, COL4A4, COL4A5, COQ2, COQ6, CRB2, CTNS, CUBN, CYP11B1, CYP11B2, DACT1, DCDC2, DGKE, DSTYK, EMP2, EYA1, FAN1, FAT1, FGA, FN1, FOXC2, FRAS1, FREM1, FREM2, GATA3, GLA, GLIS2, GRHPR, GRIP1, GSN, HNF1B, HOGA1, HPSE2, IFT27, IFT81, INF2, INVS, IQCB1, ITGA3, ITGA8, JAG1, KANK1, KANK2, KANK4, KIAA0556, KIAA0586, LAMB2, LMNA, LMX1B, LRIG2, LYZ, MAFB, MAGI2, MAP7D3, MAPKBP1, MUC1, MYH11, MYH9, MYO1E, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NUP107, NUP205, NUP93, NXF5, OCRL, OFD1, OSGEP, PAX2, PBX1, PDSS1, PDSS2, PKD1, PKD2, PKHD1, PLCE1, PMM2, PTPRO, REN, RMND1, ROBO2, RPGRIP1L, RRM2B, SALL1, SCARB2, SDCCAG8, SGPL1, SIX5, SLC41A1, SLC4A1, SLC7A7, SMARCAL1, SOX17, TBX18, TMEM67, TNXB, TRAF3IP1, TRAP1, TRPC6, TTC21B, UMOD, VIPAS39, VPS33B, WDR19, WT1, XPNPEP3, ZMPSTE24, ZNF423
Copy number analysis*: ☐ HNF1B ☐ NPHP1

☐ **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
Copy number analysis *: ☐ AVPR2

☐ **Electrolyte disorder (including Bartter syndrome, Gitelman syndrome and hypomagnesemia)*** (NEF09v18.1; 29 genes)
BSND, CACNA1S, CASR, CLCN5, CLCNKA, CLCNKB, CLDN16, CLDN19, CNNM2, DGAT1, EGF, EPCAM, FXYD2, GUCY2C, HNF1B, KCNJ1, KCNJ10, MAGED2, MYO5B, NEUROG3, PCBD1, SCN4A, SLC12A1, SLC12A3, SLC26A3, SLC41A1, SLC9A3, SPINT2, TRPM6
Copy number analysis*: ☐ CLCNKB ☐ SLC12A3

☐ **Hyperuricemia / Uricosuria** (NEF08v16.2; 14 genes)
ALDOB, ALMS1, ATP7B, CTNS, G6PC, GALT, HPRT1, PYGM, REN, SARS2, SLC22A12, SLC2A9, SLC37A4, UMOD

☐ **Renal Fanconi Syndrome** (NEF16v18.1; 32 genes)
ALDOB, AMN, ARSA, ATP7B, BCS1L, CLCN5, COQ7, COQ9, COX10, CTNS, CUBN, EHHADH, FAH, FAHD2A, G6PC, GALT, GLA, HNF4A, LRP2, OCRL, PSAP, RMND1, SLC16A12, SLC2A2, SLC2A2, SLC34A1, SLC37A4, SLC5A2, SLC6A19, SLC6A20, VIPAS39, VPS33B

☐ **Nephrocalcinosis / Nephrolithiasis*** (NEF10v18.1; 53 genes)
AGXT, ALDOB, AP2S1, APRT, ATP6V0A4, ATP6V1B1, ATP7B, BSND, CA2, CASR, CLCN5, CLCNKB, CLDN16, CLDN19, CTNS, CYP24A1, DMP1, ENPP1, FAM20A, FGF23, G6PC, GALT, GNA11, GRHPR, HNF4A, HOGA1, HPRT1, KCNJ1, KL, MAGED2, OCRL, PHEX, PTH1R, SCNN1B, SCNN1G, SLC12A1, SLC22A12, SLC2A9, SLC34A1, SLC34A3, SLC36A2, SLC37A4, SLC3A1, SLC4A1, SLC6A19, SLC6A20, SLC7A9, SLC9A3R1, TRPM6, VDR, VIPAS39, VPS33B, XDH
Copy number analysis*: ☐ SLC3A1 ☐ SLC7A9

☐ **Renal phosphate-handling** (NEF18v16.1; 8 genes)
DMP1, FGF23, FGFR1, GALNT3, PHEX, SLC34A1, SLC34A3, SLC9A3R1

☐ **Renal Tubular Acidosis (**NEF19v18.1; 17 genes)
ATP6V0A4, ATP6V1B1, BSND, CA2, CLCNKB, COQ9, EHHADH, FBXL4, FN1, G6PC, KCNJ1, SLC12A1, SLC12A3, SLC37A4, SLC4A1, SLC4A4, UQCC2

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

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[δ] Sequence and copy number analysis

^ Repeat expansion analysis only

Renal disease (Continued)
Gene panels

☐ **Hereditary kidney disease full panel** (NEF00v18.1; 380 genes including kidney tumour associated genes)
Pre-test genetic counselling required

ACE, ACTG2, ACTN4, ADAMTS13, ADCK3, ADCK4, AGT, AGTR1, AGXT, AHI1, ALDOB, ALG1, ALG8, ALMS1, AMN, ANKS3, ANKS6, ANLN, ANO1, AP2S1, APOA1, APOL1, APRT, AQP2, ARHGAP24, ARHGDIA, ARL13B, ARL6, ARSA, ATP6V0A4, ATP6V1B1, ATP7B, ATXN10, AVP, AVPR2, B2M, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCS1L, BICC1, BMP4, BMPR2, BSND, C2CD3, C3, C5orf42, CA2, CACNA1H, CACNA1S, CASR, CC2D2A, CCDC114, CD151, CD2AP, CD46, CDKN1C, CEP120, CEP164, CEP290, CEP41, CEP83, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CHD1L, CHD7, CHRM3, CLCN5, CLCNKA, CLCNKB, CLDN16, CLDN19, CNNM2, COL4A1, COL4A3, COL4A4, COL4A5, COQ2, COQ4, COQ6, COQ7, COQ9, COX10, CPT2, CRB2, CSPP1, CTNS, CUBN, CUL3, CYP11B1, CYP11B2, CYP17A1, CYP24A1, DACT1, DCDC2, DDX59, DGAT1, DGKE, DMP1, DNAJB11, DST, DSTYK, DYNC2H1, DYNC2LI1, DZIP1L, EGF, EHHADH, EMP2, ENPP1, EPCAM, EVC, EVC2, EYA1, FAH, FAHD2A, FAM134B, FAM20A, FAM58A, FAN1, FAT1, FBXL4, FGA, FGF20, FGF23, FGF8, FGFR1, FH, FLCN, FN1, FOXC2, FOXF1, FRAS1, FREM1, FREM2, FXYD2, G6PC, GALNT3, GALT, GANAB, GATA3, GDNF, GLA, GLI3, GLIS2, GLIS3, GNA11, GPC3, GPC5, GREB1L, GRHPR, GRIP1, GSN, GUCY2C, HAAO, HNF1B, HNF4A, HOGA1, HOXD13, HPRT1, HPSE2, HSD11B2, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT57, IFT80, IFT81, IKBKAP, INF2, INPP5E, INVS, IQCB1, ITGA3, ITGA8, ITGB4, JAG1, KAL1, KANK1, KANK2, KANK4, KCNJ1, KCNJ10, KCNJ5, KIAA0556, KIAA0586, KIF14, KIF7, KL, KLHL3, KYNU, LAGE3, LAMB2, LCAT, LMNA, LMOD1, LMX1B, LPP, LRIG2, LRP2, LRP4, LYZ, LZTFL1, MAFB, MAGED2, MAGI2, MAP7D3, MAPKBP1, MET, MKKS, MKS1, MUC1, MYH11, MYH9, MYO1E, MYO5B, NEK1, NEK8, NEUROG3, NGF, NOTCH2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NR3C1, NR3C2, NUP107, NUP205, NUP93, NXF5, OCRL, OFD1, OSGEP, PAX2, PAX8, PBX1, PCBD1, PDE6D, PDSS1, PDSS2, PHEX, PKD1, PKD2, PKHD1, PLCE1, PMM2, PODXL, PRDM12, PRKCSH, PSAP, PTEN, PTH1R, PTPRO, PYGM, REN, RET, RMND1, ROBO2, RPGRIP1, RPGRIP1L, RRM2B, SALL1, SALL4, SARS2, SCARB2, SCLT1, SCN11A, SCN4A, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SDHB, SEC61A1, SEC61B, SEC63, SGPL1, SIX1, SIX2, SIX5, SLC12A1, SLC12A3, SLC16A12, SLC22A12, SLC26A3, SLC2A2, SLC2A9, SLC34A1, SLC34A3, SLC36A2, SLC37A4, SLC3A1, SLC41A1, SLC4A1, SLC4A4, SLC5A2, SLC6A19, SLC6A20, SLC7A7, SLC7A9, SLC9A3, SLC9A3R1, SLIT2, SMARCAL1, SMARCAL1, SOX17, SPINT2, SPTLC1, SPTLC2, STRA6, STX16, TBC1D1, TBX18, TCTEX1D2, TCTN1, TCTN2, TCTN3, THBD, TMEM104, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TNXB, TP53RK, TPRKB, TRAF3IP1, TRAP1, TRIM32, TRPC6, TRPM6, TSC1, TSC2, TTC21B, TTC8, UMOD, UPK3A, UQCC2, VDR, VHL, VIPAS39, VPS33B, WDPCP, WDR19, WDR34, WDR35, WDR60, WDR73, WNK1, WNK4, WNT4, WT1, XDH, XPNPEP3, XPO5, YRDC, ZEB2, ZIC3, ZMPSTE24, ZNF423

☐ **Hypertension / Pseudohypoaldosteronism*** (NEF15v18.1; 18 genes)

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

Copy number analysis*: ☐ WNK1

Renal disease
Single gene | Sequence analysis

☐ Atypical hemolytic uremic syndrome 1 (AHUS1)[δ]	CFH[δ]
☐ Atypical hemolytic uremic syndrome 2 (AHUS2)[δ]	CD46[δ]
☐ Atypical hemolytic uremic syndrome 3 (AHUS3)[δ]	CFI[δ]
☐ Branchiootorenal syndrome 1 (BOR1)[δ]	EYA1[δ]
☐ Branchiootorenal syndrome 2 (BOR2)	SIX5
☐ Branchiootorenal syndrome 3 (BOS3)	SIX1
☐ Branchiootic syndrome (BOS1)	EYA1
☐ Familiar vesicoureteral reflux (VUR2)[δ]	ROBO2
☐ Focal segmental glomerulosclerosis 1 (FSGS1)	ACTN4
☐ Focal segmental glomerulosclerosis 2 (FSGS2)	TRPC6
☐ Focal segmental glomerulosclerosis 3 (FSGS3)	CD2AP
☐ Focal segmental glomerulosclerosis 5 (FSGS5)	INF2
☐ Gitelman syndrome[δ]	SLC12A3[δ]
☐ Glomerulopathy with fibronectin deposition (GFND2) *[NEF06v16.1]*	FN1
☐ Hirschsprung disease 3, susceptibilty to (HSCR3)	GDNF
☐ Hypertension and brachydactyly syndrome/Bilginturan syndrome	PDE3A
☐ Hypoparathyroidism, sensorineural deafness, and renal dysplasia	GATA3
☐ Interstitial lung disease, nephrotic syndrome	ITGA3
☐ Joubert syndrome type 3 (JBTS3)	AHI1
☐ Joubert syndrome type 4 (JBTS4)[δ]	NPHP1[δ]
☐ Joubert syndrome type 12 (JBTS12)	KIF7
☐ Nephronophthisis 1[δ]	NPHP1[δ]
☐ Nephronophthisis 3	NPHP3
☐ (Nephrogenic) diabetes insipidus	AQP2
☐ (Nephrogenic) central diabetes insipidus	AVP
☐ (Nephrogenic) X- linked diabetes insipidus[δ]	AVPR2[δ]
☐ Nephrotic syndrome, congenital Finnish type (NPHS1)	NPHS1
☐ Nephrotic syndrome, steroid resistant (NPHS2)	NPHS2
☐ Nephrotic syndrome type 3, early onset (NPHS3)	PLCE1
☐ Nephrotic syndrome met diffuse mesangial sclerosis, (NPHS4)	WT1
☐ Pierson syndrome, congenital	LAMB2
☐ Papillorenal syndrome	PAX2
☐ Renal adysplasia[δ]	RET[δ]
☐ Renal adysplasia	UPK3A
☐ Renal cysts and diabetes syndrome[δ]	HNF1B[δ]

Other diseases
Gene panels

☐ **Congenital diarrhoea** (DIA00v17.1; 64 genes)

ADA, ADAM17, AIRE, ANGPTL3, ANKZF1, APOB, CD3D, CD3E, CFTR, CLMP, DCLRE1C, DGAT1, EPCAM, FLNA, FOXP3, GUCY2C, IL10, IL10RA, IL10RB, IL12RB1, IL21, IL2RA, IL2RG, IL7R, JAK3, LCT, MPI, MTTP, MYO5B, NCF4, NEUROG3, NHEJ1, NPC1L1, PCSK1, PCSK9, PNLIP, PNP, PRSS1, PTPRC, RAG1, RAG2, SAR1B, SBDS, SI, SKIV2L, SLC10A2, SLC26A3, SLC2A2, SLC39A4, SLC5A1, SLC7A7, SLC9A, SPINK1, SPINT2, STAT1, STAT5B, STX3, TCN2, TMPRSS15, TTC37, TTC7A, UBR1, XIAP, ZAP70

☐ **Hereditary angioedema, broad differential diagnosis** (HAE00v18.1; 51 genes)

A2M, ACE, ANGPT1, BDKRB1, BDKRB2, CPB2, CPM, CPN1, CPN2, DPP4, F11, F12, F13B, F2, HRH1, HRH3, HRH4, KLK1, KLK10, KLK11, KLK12, KLK13, KLK14, KLK15, KLK2, KLK3, KLK4, KLK5, KLK6, KLK7, KLK8, KLK9, KLKB1, KNG1, MASP1, MASP2, PLAU, PLAUR, PLG, PTGS1, PTGS2, SERPINA1, SERPINA4, SERPINB2, SERPINE1, SERPINF2, SERPING1, TFPI, VEGFA, XPNPEP1, XPNPEP2

☐ **Hereditary angioedema** (HAE01v18.1; 4 genes)

ANGPT1, F12, PLG, SERPING1

☐ **Familial partial lipodystrophy (FPLD) and congenital generalized lipodystrophy (CGL)** (LIP01v17.1; 9 genes)

PPARG, LMNA, CIDEC, AKT2, AGPAT2, BSCL2, CAV1, PTRF, ZMPSTE24

☐ **Idiopathic pulmonary fibrosis** (IPF01v19.1; 24 genen)

ABCA3, AP3B1, ASAH1, CSF2RA, CSF2RB, DKC1, FAM111B, GBA, HPS1, HPS4, ITGA3, NKX2-1, NOP10, PARN, RTEL1, SFTPA2, SFTPB, SFTPC, SLC34A2, SLC7A7, SMPD1, TERC, TERT, TINF2

☐ **Neonatal erythroderma** (ERY00v17.1; 60 genes)

ABCA12, ABHD5, ADAM17, ALDH3A2, ALOX12B, ALOXE3, ASS1, ATP7A, BCKDHA, BCKDHB, BTD, BTK, C5, C8A, C8B, C8G, CARD14, CLDN1, CPS1, CYP4F22, CERS3, CDSN, DCLRE1C, DSG1, DBT, DLD, EBP, ELOVL4, ERCC2, ERCC3, GBA, GJB2, GJB6, GTF2H5, HLCS, IL36RN, KIT, KRT1, KRT10, KRT2, LIPN, LOR, MPLKIP, MBTPS2, MUT, NIPAL4, NSDHL, PCCA, POMP, PNPLA1, PCCB, RAG1, RAG2, STST, SLC25A13, SLC30A2, SLC39A4, SPINK5, TBX1, TGM1

☐ **Nonsyndromal disorders of sex development (DSD)** (DSD00v16.1; 32 genes)

AMH, AMHR2, AR, CBX2, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, DHH, DMRT1, HSD17B3, HSD3B2, LHB, LHCGR, MAMLD1, MAP3K1, NR0B1, NR3C1, NR5A1, POR, PSMC3IP, RSPO1, SOX3, SOX9, SRD5A2, SRY, STAR, TSPYL1, WNT4, WT1, ZFPM2

* NGS gene panel analysis can only detect single nucleotide changes and small deletions/duplications. Large copy number changes and repeat expansions cannot be detected. Unless indicated otherwise, these analyses must be requested seperately.

[δ] Sequence and copy number analysis

^ Repeat expansion analysis only

Other diseases
Single gene | Sequence analysis

- ☐ Azoospermia, severe oligozoospermia (AZF) *(Copy number analysis only)* [AZF]
- ☐ Adrenal hypoplasia, X-linked, (AHC)[δ] NR0B1[δ]
- ☐ Fragile X-associated tremor/ataxia syndrome (FXTAS)^ FMR1^
- ☐ Microvillus inclusion disease (MVID) or Diarrhea 2, with microvillus atrophy (DIAR2)[δ] MYO5B[δ]
- ☐ Gonadal dysgenesis, partial or complete, with or without renal failure, (POF7) NR5A1
- ☐ Persistent Mullerian duct syndrome, (PMDS), type 1 AMH
- ☐ Persistent Mullerian duct syndrome, (PMDS), type 2 AMHR2
- ☐ Premature ovarian failure, (POF1)^ FMR1^
- ☐ Surfactant metabolism dysfunction, pulmonary 3 (SMDP3) ABCA3

- ☐ Uniparental disomy, chromosome:…… [MARK]
- ☐ X-chromosome inactivation AR
- ☐ 15q11-q13 duplication syndrome *(methylation specific copy number analysis)* [15q11-q13]

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[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 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**.