

Department of Medical Genetics

REQUEST FORM

Section Genome Diagnostic

Universitair Medisch Centrum Utrecht
Centrale Balie CDL
Huispost G.03.3.30
Heidelberglaan 100
3584 CX Utrecht



UMC Utrecht

PERSONAL DETAILS (please complete in capitals)

Family name + initials

address
zip code + residence
country

date of birth

gender

billing details

Secretary

Tel 088 – 75 54090

Fax 088 – 75 55034

E-mail genoomdiagnostiek@umcutrecht.nl

REFERRING PHYSICIAN (please complete in captials)

Naam :

Hospital :

Address :

Zip code/residence :

Country :

Date :

Your reference :

copy results :

Telephone :

E-mailaddress :

INDICATION

Please indicate the desired gene panel analyses, deletion test (MLPA) or individual gene analysis (see table next pages).
Please also include relevant clinical data and/or family pedigree. Use only one form for each patient!

MATERIAL

Counselor does not grant permission for anonymous use of leftover material (see last page: use of patient material).

- No permission to use leftover material

Tubes of blood (or DNA samples) please label clearly with name, gender and date of birth.

Urgent and prenatal requests: only after consultation (tel. +31 (0)88-7554090)

- Blood (2 x 10 ml EDTA, 2 x 3 ml for neonates)
 Chorionic villi
 Amniotic fluid
 Blood for RNA isolation (2 x 2,5 ml PAXgene blood tubes) (only after consultation)
 DNA number(s)
 Tissue type
 number of sample(s)

PURPOSE

- Confirmation of clinical diagnosis / exclusion of a diagnosis
 Carriership (known gene defect in the family)
 Presymptomatic testing
 Partner testing
 Prenatal testing (only after consultation)
 Archiving (for possible future diagnosis)
 Research
 URGENT (only after consultation)

FAMILY HISTORY

- Mutation unknown
 Mutation known
 (relation to index patient given in family tree on reverse of form)

Gene:

Mutation:

Family number:

Reference:

To be completed by laboratory

ETIKETTEN

REGISTRATIE

U-nummer

--	--	--	--	--	--	--

Datum::

Indicatie:

Gericht / Volledig

Paraaf:

Ontvangstdatum:

Cardiovascular disease

- Cardiomyopathy (62 genes)
 - 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, DES, DMD, DSC2, DSG2, DSP, DTNA, EMD, EYA4, FHL1, FLNC, FKTN, GATAD1, GLA, ILK, JPH2, JUP, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK, MYLK2, MYOT, MYOZ1, MYOZ2, MYPN, NEBL, NEXN, PDLIM3, PKP2, PLN, PRKAG2, RBM20, RYR2, SCN5A, SGCD, TAZ, TCAP, TMEM43, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTR, VCL*

Deletion test MYBPC3 PKP2

- Conduction abnormalities (33 genes)
 - Sudden cardiac arrest
 - Sudden unexplained death
 - Arrhythmogenic right ventricle (ARVD/C)
 - Brugada syndrome (BrS)
 - Sick Sinus syndrome (SSS)
 - Atrial standstill
 - Catecholaminergic polymorphic VT's (CPVT)
- AKAP9, ANK2, CACNA1C, CACNA2D1, CACNB2, CASQ2, CAV3, DES, 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*

Deletion test PKP2 KCNQ1/KCNH2

- Congenital heart defects (35 genes)
 - Non-syndromal
 - ASD/VSD/DORV
 - Heterotaxy
 - Tetralogy of Fallot (TOF)
 - Syndromal
 - Velocardiofacial/DiGeorge (DGS)
 - Oculo-Facio-Cardio Dental
 - Holt-Oram (HOS)
 - Alstrom (ALMS)
 - Alagille (AGS)
 - Wolff-Parkinson-White (WPW)

Cardiovascular disease (continued)

- Cantu
 - Noonan/LEOPARD (NS/LS)
 - Cardio-Facio-Cutaneous (CFC)
 - ALMS1, ACTC1, ACVR2B, BRAF, CBL, CFC1, CRELD1, ELN, FOXH1, GATA4, GDF1, GJA1, GJC1, HRAS, JAG1, KRAS, LDB3, LEFTY2, MAP2K1, MAP2K2, MYBPC3, MYH11, MYH6, MYH7, NKX2-5, NODAL, NRAS, PTPN11, RAF1, SHOC2, SOS1, TAZ, TBX20, TBX5, ZIC3*
- Deletion test MYBPC3 JAG1
- Analysis Titin gene
TTN
 - Vascular disorders (17 genes)
 - Familial thoracic aortic aneurysm and aortic dissection (TAAD)
 - Marfan (MFS)
 - Loays-Dietz (LDS)
 - ACTA2, CBS, COL3A1, COL5A1, COL5A2, ELN, FBN1, FBN2, MYH11, MYLK, SLC2A10, SLC8A1, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2*

Dysmorphology

- Fraser syndrome (4 genes)
FRAS1, FREM2, FREM1, GRIP1

Epilepsy

- Benign neonatal/infantile convulsions (5 genes)
KCNQ2, KCNQ3, PRRT2, SCN2A, TBC1D24
 - Deletion test KCNQ2 KCNQ3
- Epileptic encephalopathy (EIEE) (32 genes)
ALDH7A, ARHGEF9, ARX, CDKL5, FOXG1, GRIN2A, GRIN2B, KCNQ2, KCNT1, MAGI2, MAPK10, MECP2, MEF2C, PCDH19, PLCB1, PNKP, PNPO, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SLC19A3, SLC25A22, SLC2A1, SPTAN1, SRGAP2, STXBP1, SYNGAP1, TBCE, UBE3A, ZEB2
 - Deletion test ARX CDKL5 FOXG1 KCNQ2
 - MECP2 MEF2C PCDH19 SCN1A SLC2A1
- Febrile seizures / Genetic epilepsy with febrile seizures plus (GEFS+) (10 genes)
CLCN2, GABRD, GABRG2, GPR98, PCDH19, SCN1A, SCN1B, SCN2A, SCN9A, TBC1D24
 - Deletion test PCDH19 SCN1A
- Focal epilepsy (9 genes)
CHRNA2, CHRNA4, CHRN2, ELP4, GRIN2A, KCNT1, LGI1, SRPX2, SYN1
 - Deletion test CHRNA4 CHRN2

Epilepsy (continued)

- Progressive myoclonic epilepsy (8 genes)
ASAH1, CSTB, EPM2A, KCTD7, NHLRC1, PRICKLE1, PRICKLE2, SCARB2
- Deletion test EPM2A NHLRC1
- Metabolic disease with epilepsy (24 genes)
ADSL, ALDH7A1, AMT, CLN3, CLN5, CLN6, CLN8, CPT2, CTSD, DNAJC5, FOLR1, GAMT, GCSH, GLDC, GLRA1, GLRB, GPHN, MFSD8, MTHFR, PNPO, PPT1, PPT2, SLC2A1, TPP1
- Deletion test GLDC SLC2A1
- IGE/JME/CAE (11 genes)
BRD2, CACNA1H, CACNB4, CASR, CLCN2, EFHC1, GABRA1, GABRB3, GABRD, ME2, SLC2A1
- Deletion test SLC2A1
- Epilepsy with paroxysmal disorders (5 genes)
ATP1A2, CACNA1A, KCNMA1, PRRT2, SLC2A1
- Deletion test SLC2A1
- Epileptic syndromes with epilepsy and intellectual disability (50 genes)
ARX, ATP6AP2, ATRX, AUTS2, CASK, CDKL5, CNKSR2, CNTNAP2, CUL4B, DCX, DYRK1A, FGD1, FOXP1, GPC3, GRIA3, GRIN2A, GRIN2B, HSD17B10, KCNJ10, KDM5C, MBD5, MECP2, MED12, MEF2C, NRXN1, OFD1, OPHN1, PAK3, PHF6, PLP1, PNKP, PQBP1, RAB39B, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SLC6A8, SLC9A6, SMC1A, SMS, SRPX2, STXBP1, SYNGAP1, SYP, TBC1D24, TCF4, TREX1, UBE3A, ZEB2
- Deletion test ARX CDKL5 FOXP1 MECP2
MEF2C NRXN1
- Inflammatory epilepsy (2 genes)
RANBP2, SCN1A
- Deletion test SCN1A

Hereditary tumors

- Breast cancer (2 genes)
BRCA1, BRCA2
- Deletion test BRCA1 BRCA2

Metabolic diseases

- Glycine encephalopathy/non-ketonic hyperglycinemia (3 genes)
AMT, GCSH, GLDC
- Intrahepatic cholestasis (3 genes)
ATP8B1, ABCB11, ABCB4
- Serine synthesis defect (3 genes)
PHGDH, PSPH, PSAT1

Neuromuscular diseases

- Nemaline myopathy (NEM) (7 genes)
ACTA1, CFL2, KBTBD13, NEB, TPM2, TPM3, TNNT1
- Myofibrillaire myopathy (MFM) (7 genes)
BAG3, CRYAB, DES, FHL1, FLNC, LDB3, MYOT
- Minicore myopathy/ Central core disease
RYR1

Neurological diseases

- FTD-ALS (15 genes)
ALS2, ANG, CHMP2B, FIG4, FUS, GRN, MAPT, NPC1, NPC2, SETX, SMPD1, SOD1, TARDBP, VAPB, VCP
- Repeat expansion analysis C9ORF72
- Ataxia (AD + AR ataxia) (16 genes)
AFG3L2, APTX, AR, ATXN1, ATXN2, ATXN3, ATXN7, CACNA1A, FGF14, FXN, KCNA1, KCNC3, PDYN, PRKCG, SETX, TBP
- Repeat expansion analysis ATX1 ATX2 ATX3
ATX7 CACNA1A PPP2R2B TPB FMR1 (FXTAS)
- Cerebral cavernous malformations (CCM) (3 genes)
KRIT1, CCM2, PDCD10
- Deletion test KRIT1 CCM2 PPDCD10

Primary immunodeficiencies

- Complete gene panel (184 genes)
ACP5, ACTB, ADA, AICDA, AIRE, AK2, AP3B1, ATM, ATP2A2, BLM, BLNK, BTK, BTLA, C1QA, C1QB, C1R, C1S, C2, C5, C3, C6, C7, C8A, C8B, C9, CASP10, CASP8, CBL, CD19, CD27, CD3D, etc. see www.umcutrecht.nl/erfelijkheid
- Autoinflammatory (19 genes)
IL1RN, LPIN2, MEFV, MVK, NLRP12, NLRP3, NLRP7, NOD2, PSTPIP1, NFRSF1A, PSENEN, NCSTN, IL36RN, IL10, IL10RA, IL10RB, PLCG2, PSMB8, SH3BP2
- Deletion test PLCG2 IL1RN IL10RB
- HLH/Immune dysregulation (9 genes)
PRF1, UNC13D, STX11, STXBP, SH2D1A, XIAP, LYST, RAB27A, AP3B
- Deletion test PRF1 UNC13D STX11
- ALPS/Autoimmunity (12 genes)
FAS, FASLG, CASP10, CASP8, KRAS, NRAS, FADD, AIRE, FOXP3, IL2RA, ITCH, LRBA
- Deletion test PRF1 UNC13D STX11

Primary immunodeficiencies (continued)

(S)CID (28 genes)

ADA, AK2, CD3D, CD3E, CD3G, CD40, CD8A, CORO1A, DCLRE1C, IL2RA, IL2RG, IL7RA, JAK3, LIG4, NHEJ1, PNP, PRKDC, PTPRC, RAG1, RAG2, ZAP70, CD40LG, PTPRC, ORAI1, STIM1, STAT5B, DOCK8, TBX1

Deletion test DOCK8

B cell pathology (14 genes)

BTK, ICOS, CD19, CD81, TNFRSF13B, TNFRSF13C, CD40, CD40L, AICDA, UNG, CD79A, BLNK, CD79B, IGLL1

HIES syndromes (3 genes)

STAT3, TYK2, DOCK8

Deletion test DOCK8

CMC (6 genes)

IL17RA, IL17F, STAT1, TLR3, AIRE, IL2RA

Deletion test DOCK8

Other gene panels

Erythrocyte membrane disorders (7 genes)

SPTA1, SPTB, ANK1, SLC4A1, EPB41, EPB42, RHAG

Deletion test SPTA1 SPTB ANK1

SLC4A1 EPB41, EPB42, RHAG

Obesitas (6 genes)

ALMS1, LEP, LEPR, PCSK1, POMC, MC4R,

Amelogenesis imperfecta (9 genes)

AMELX, C4orf26, DLX3, ENAM, FAM20A, FAM83H, KLK4, MMP20, WDR72



The section genome diagnostics has been certified with NEN-EN-ISO 15189:2007 by the Accreditation Council. The scope of accreditation number M001 can be seen on www.rva.nl.

Cardiovascular diseases

o Alstrom syndrome	ALMS1
o Alveolar capillary dysplasia with misalignment of the pulmonary veins, ACDMPV	FOXF1
o Arrhythmogenic right ventricular dysplasia, ARVD/C1	TGFB3
o Arrhythmogenic right ventricular dysplasia, ARVD/C5	TMEM43
o Arrhythmogenic right ventricular dysplasia, ARVD/C8	DSP
o Arrhythmogenic right ventricular dysplasia, ARVD/C9	PKP2
o Arrhythmogenic right ventricular dysplasia, ARVD/C10	DSG2
o Arrhythmogenic right ventricular dysplasia, ARVD/C11	DSC2
o Arrhythmogenic right ventricular dysplasia, ARVD/C12	JUP
o Arrhythmogenic right ventricular dysplasia, ARVD/C	DES
o Arrhythmogenic right ventricular dysplasia, ARVD/C	PLN
o Arrhythmogenic right ventricular dysplasia, ARVD/C	LMNA
o Brugada syndrome	SCN1B
o Cantú syndrome	ABCC9
o Cardiomyopathy, dilated, DCM	LMNA
o Cardiomyopathy, dilated, DCM	DES
o Cardiomyopathy, dilated and cataract, DCM	CRYAB
o Cardiomyopathy, dilated, hypertrophic, DCM/HCM	TNNT2
o Cardiomyopathy, dilated, hypertrophic, DCM/HCM	PLN
o Cardiomyopathy, dilated, hypertrophic, DCM/HCM	MYL2
o Cardiomyopathy, dilated, hypertrophic, DCM/HCM	MYLK2
o Cardiomyopathy, dilated, hypertrophic, DCM/HCM	MYOZ2
o Cardiomyopathy, dilated, hypertrophic, DCM/HCM	MYH7
o Cardiomyopathy, dilated, hypertrophic, DCM/HCM	MYBPC3
o Cardiomyopathy, dilated, hypertrophic, DCM/HCM	CASQ2
o Cardiomyopathy, dilated, hypertrophic, DCM/HCM	CAV3
o Cardiomyopathy, dilated, hypertrophic, DCM/HCM	FHL1
o Cardiomyopathy, dilated, hypertrophic, DCM/HCM	TCAP
o Cardiomyopathy, dilated, hypertrophic, DCM/HCM	TNNC1
o Cardiomyopathy, dilated, hypertrophic, DCM/HCM	TNNI3
o Cardiomyopathy, dilated, hypertrophic, DCM/HCM	TPM1
o Cardiomyopathy, dilated, hypertrophic, DCM/HCM	TTN
o Cataract and dilated cardiomyopathy	CRYAB
o Fabry disease, alpha-galactosidase A deficiency	GLA
o Fallot, Tetralogy of, TOF	NKX2-5
o Holt-Oram syndrome, HOS	TBX5
o Cardiomyopathy and amyloid polyneuropathy	TTR
o Oculofaciocardiodental syndrome, OFCD	BCOR
o Syndromal microphthalmia 2, MCOPS2	BCOR
o Velocardiofacial syndrome (VCF)/DiGeorge Syndrome	TBX1
o Ventricular tachycardia, catecholaminergic polymorphic type 2, CPVT2	CASQ2

Dysmorphology

o Acrocallosaal Syndrome, ACLS	KIF7
o Albright hereditary osteodystrophy, AHO	GNAS
o Amelogenesis imperfecta A14	DLX3
o Cantú syndrome	ABCC9
o Cleidocranial dysplasia, CCD	RUNX2
o Currarino syndrome, TRIAD	MXN1
o Floating-Harbor Syndrome, FHS	SRCAP
o Fraser syndrome	FRAS1
o Fraser syndrome	FREM2
o Fraser syndrome / MOTA / BNAR / Trigonocephaly 2	FREM1
o Fraser syndrome	GRIP1
o Hypodontia, HYD1	MSX1
o Hypodontia, HYD3	PAX9
o Hypodontia	AXIN2
o Hypodontia	IRF6
o Hypodontia	ITM2A
o Hypodontia	SUMO1
o Hypodontia	TBX22
o Hypodontia	WNT10A
o McCune-Albright syndrome, MAS	GNAS
o Microphthalmia, syndromic type 2, MCOPS2	BCOR
o Oculofaciocardiodental syndrome, OFCD	BCOR
o Osseous heteroplasia progressive, POH	GNAS
o Trichodontoosseous syndrome, TDO	DLX3
o Van der Woude syndrome	IRF6

Epilepsy

o Autosomal dominant lateral temporallobe – epilepsy ADLTE	LGI1
o Benign familial infantile seizures type 2, BFIS2	PRRT2
o Benign familial neonatal seizures, BFNC	KCNQ2
o Benign familial neonatal seizures, BFNC	KCNQ3

Epilepsy (continued)

o Benign familial neonatal-infantile seizures, BFNIS	SCN2A
o Cortical dysplasia-focal epilepsy syndrome, CDFE	CNTNAP2
o Dravet syndrome SMEI/SMEB	SCN1A
o Early infantile epileptic encephalopathy type 1, EIEE1	ARX
o Early infantile epileptic encephalopathy type 2, EIEE2	CDKL5
o Early infantile epileptic encephalopathy type 3, EIEE3	SLC25A22
o Early infantile epileptic encephalopathy type 4, EIEE4	STXBP1
o Early infantile epileptic encephalopathy type 7, EIEE7	KCNQ2
o Early infantile epileptic encephalopathy type 8, EIEE8	ARHGEF9
o Early infantile epileptic encephalopathy type 9, EIEE9	PCDH19
o Early infantile epileptic encephalopathy type 10, EIEE10	PNKP
o Early infantile epileptic encephalopathy type 11, EIEE11	SCN2A
o Early infantile epileptic encephalopathy type 12, EIEE12	PLCB1
o Genetic epilepsy with febrile seizures plus, GEFS+	SCN1A
o Genetic epilepsy with febrile seizures plus, GEFS+	SCN1B
o Genetic epilepsy with febrile seizures plus, GEFS+	SCN2A
o Genetic epilepsy with febrile seizures plus, GEFS+	GABRG2
o GLUT1 deficiency syndrome type 1 and - 2, GLUT1DS1/GLUT1DS2	SLC2A1
o Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations	MEF2C
o Nocturnal frontal lobe epilepsy type 1, ADNFLE1	CHRNA4
o Nocturnal frontal lobe epilepsy type 3, ADNFLE3	CHRN2
o Progressive myoclonic epilepsy type 1A, EPM1	CSTB
o Progressive myoclonic epilepsy type 1B, EPM1B	PRICKLE1
o Progressive myoclonic epilepsy type 2A, EMP2A / Lafora	EPM2A
o Progressive myoclonic epilepsy type 2B, EPM2B / Lafora	NHLRC1
o Progressive myoclonic epilepsy type 3, EPM3	KCTD7
o Progressive myoclonic epilepsy type 4, AMRF, EPM4	SCARB2
o Progressive myoclonic epilepsy type 5, EPM5	PRICKLE2
o Progressive myoclonic epilepsy type 6, EPM6	GOSR2
o Pyridoxine-dependent epilepsy, PDE	ALDH7A1
o Pyridoxine-dependent epilepsy, PDE	PNPO
o Unverricht-Lundborg disease, UL	CSTB
o X-linked Rolandic epilepsy, mental retardation and speech dyspraxia, RESDX	SRPX2

Hereditary tumors

o Acromegalia. Pituitary adenoma predisposition, PAP	AIP
o Breast-ovarian cancer, familial	BRCA1
o Breast-ovarian cancer, familial	BRCA2
o Colorectal-oligodontia cancer syndrome	AXIN2
o Emberger syndrome	GATA2
o Familial acute myeloid leukemia, AML	CEBPA
o Familial acute myeloid leukemia / platelet disorder, AML/FDP	RUNX1
o Hyperparathyroidism, familial primary, HRPT1	MEN1
o Lynch syndrome, HNPCC1	MSH2
o Lynch syndrome, HNPCC5	MSH6
o Multiple endocrine neoplasia type 1, MEN1	MEN1
o Multiple endocrine neoplasia type 2A, MEN2A	RET
o Multiple endocrine neoplasia type 4 / atypical	CDKN1A
o Multiple endocrine neoplasia type 4 / atypical	CDKN1B
o Multiple endocrine neoplasia type 4 / atypical	CDKN2B
o Multiple endocrine neoplasia type 4 / atypical	CDKN2C
o Ovarian carcinoma, hereditary	BRIP1
o Papillary renal cell carcinoma, familial, PRC	MET
o Pheochromocytoma / paraganglioma, FEO/PGL	SDHB
o Pheochromocytoma / paraganglioma, FEO/PGL	SDHC
o Pheochromocytoma / paraganglioma, FEO/PGL	SDHD
o Pheochromocytoma / paraganglioma, FEO/PGL	TMEM127
o Pheochromocytoma / paraganglioma, FEO/PGL	MAX
o Pseudohyperparathyroidism type 1a, PHP1a	GNAS
o Sporadic medullary thyroid carcinoma, MTC	RET
o Von Hippel-Lindau syndrome	VHL

Metabolic disease

o Acyl-CoA dehydrogenase, medium chain, deficiency of	ACADM
o Biotinidase deficiency	BTD
o Cholestasis, intrahepatic type 1, BRIC/PFIC type 1	ATP8B1
o Cholestasis, intrahepatic type 2, BRIC/PFIC type 2	ABCB11
o Cholestasis, intrahepatic type 3, BRIC/PFIC type 3	ABCB4
o Congenital disorder of glycosylation type 1a, CDG1A	PMM2
o Congenital disorder of glycosylation type 1P, CDG1p	ALG11

Metabolic disease (continued)

- o Fabry disease, alpha-galactosidase A deficiency
- o Glycerol kinase deficiency, GK
- o Glycine encephalopathy / nonketotic hyperglycinemia
- o Glycine encephalopathy / nonketotic hyperglycinemia
- o Glycine encephalopathy / nonketotic hyperglycinemia
- o Hartnup disorder
- o Hyperinsulinemic hypoglycemia type 7, HHF7
- o Metachromatic leukodystrophy, MLD
- o Methylmalonic aciduria type cblA
- o Phenylketonuria type 1, PKU
- o Phenylketonuria, hyperphenylalaninemia, BH4-deficient, PTPS
- o Pompe disease, Glycogen storage disease II, GSD2
- o Pyruvate kinase deficiency, PK
- o Serine biosynthesis defect, PHGDH deficiency
- o Serine biosynthesis defect, PSPH deficiency
- o Serine biosynthesis defect, PSAT1 deficiency
- o Tyrosinemia, type I
- o Wilson disease, WD

GLA
GK
AMT
GCSH
GLDC
SLC6A19
SLC16A1
ARSA
MMAA
PAH
PTS

GAA
PKLR
PHGDH
PSPH
PSAT1
FAH
ATP7B

Neurological disease

- o Amyloidosis I and VII; transthyretin amyloidosis
- o Amyotrophic lateral sclerosis type 1, ALS1
- o Amyotrophic lateral sclerosis (Juveniel) type 2, ALS2
- o Amyotrophic lateral sclerosis type 4, ALS4
- o Amyotrophic lateral sclerosis type 6, ALS6
- o Amyotrophic lateral sclerosis type 8, ALS8
- o Amyotrophic lateral sclerosis type 9, ALS9
- o Amyotrophic lateral sclerosis type 10, ALS10
- o Amyotrophic lateral sclerosis type 11, ALS11
- o Amyotrophic lateral sclerosis type 14, ALS14
- o Amyotrophic lateral sclerosis type 15, with or without FTD, ALS15
- o Amyotrophic lateral sclerosis/Frontotemporal dementia FTDLALS
- o Cerebral cavernous malformations type 1, CCM1
- o Cerebral cavernous malformations type 2, CCM2
- o Cerebral cavernous malformations type 3, CCM3
- o Frontotemporal dementia, FTD
- o Frontotemporal dementia, FTD
- o Fuhrmann syndrome
- o Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia
- o Pitt Hopkins-like syndrome 1
- o Pitt Hopkins-like syndrome 2
- o Schizencephaly
- o Spinocerebellar ataxia type 1, SCA1
- o Spinocerebellar ataxia type 2, SCA2
- o Spinocerebellar ataxia type 3, SCA3
- o Spinocerebellar ataxia type 6, SCA6
- o Spinocerebellar ataxia type 7, SCA7
- o Spinocerebellar ataxia type 12, SCA12
- o Spinocerebellar ataxia type 13, SCA13
- o Spinocerebellar ataxia type 14, SCA14
- o Spinocerebellar ataxia type 17, SCA17
- o Spinocerebellar ataxia type 23, SCA23
- o Spinocerebellar ataxia type 28, SCA28

TTR
SOD1
ALS2
SETX
FUS
VAPB
ANG
TARDBP
FIG4
VCP
UBQLN2

C9ORF72

KRIT1
CCM2
PDCD10
MAPT
GRN
WNT7A
VCP

CNTNAP2
NRXN1
EMX2
ATXN1
ATXN2
ATXN3
CACNA1A
ATXN7
PPP2R2B
KCNC3
PRKCG
TBP
PDYN
AFG3L2

Neuromuscular disease

- o Ehlers-Danlos syndrome (musculocontractural)
- o Minicore myopathy/ Central core disease
- o Muscular dystrophy, Emery-Dreifuss type 6, EDMD6
- o Muscular dystrophy, Limb-Girdle type 2G, LGMD2G
- o Myotonic dystrophy type 1, DM1
- o Myotonic dystrophy type 2, DM2
- o Myofibrillar myopathy type 1, MFM1
- o Myofibrillar myopathy type 2, MFM2
- o Nemaline myopathy type 1, NEM1
- o Nemaline myopathy type 3, NEM3
- o Nemaline myopathy type 4, NEM4
- o Nemaline myopathy type 5, NEM5
- o Nemaline myopathy type 6, NEM6
- o Nemaline myopathy type 7, NEM7
- o Spinal Muscular Atrophy (SMA type 1 - 4)

CHST14
RYR1
FHL1
TCAP
DMPK
CNBP
DES
CRYAB
TPM3
ACTA1
TPM2
TNNT1
KBTBD13
CFL2
SMN1

Obesity

- o Alstrom syndrome
- o Cohen syndrome
- o Leptin deficiency
- o Leptin receptor deficiency
- o Obesity with impaired prohormone processing
- o Proopiomelanocortin deficiency
- o Obesity, autosomal dominant

ALMS1
VPS13B
LEP
LEPR
PCSK1
POMC
MC4R

Primary immunodeficiencies

- o Acne inversa, familial type 1
- o Acne inversa, familial type 2
- o Agammaglobulinemia, X-linked, XLA
- o Autoimmune lymphoproliferative syndrome, ALPS type 1a
- o Autoimmune lymphoproliferative syndrome, ALPS type 1b
- o Autoimmune lymphoproliferative syndrome, ALPS type 2a
- o Autoimmune polyendocrinopathy syndrome, type I, APS1
- o Blau syndrome
- o Candidiasis, familial type 2
- o Candidiasis, familial type 5
- o Candidiasis, familial type 6
- o Candidiasis, familial type 7
- o CINCA syndrome
- o Cold-induced autoinflammatory syndrome FCAS1
- o Cold-induced autoinflammatory syndrome FCAS2
- o Cold-induced autoinflammatory syndrome FCAS3
- o DIRA syndrome
- o Familial Mediterranean fever, FMF
- o Hemophagocytic lymphohistiocytosis, HLH type 2
- o Hemophagocytic lymphohistiocytosis, HLH type 3
- o Hemophagocytic lymphohistiocytosis, HLH type 4
- o Hemophagocytic lymphohistiocytosis, HLH type 5
- o Hydatidiform mole, recurrent type 1
- o Hyper-IgM syndrome, CD40 ligand deficiency
- o Hyper-IgM syndrome, AID deficiency
- o Hereditaire Angioderma type 1
- o Hyper-IgE syndrome

NCSTN
PSENN
BTB
FAS

FASL

CASP10

AIRE

NOD2
CARD9
IL17RA
IL17F
STAT1
NLRP3
NLRP3
NLRP12
PLCG2
IL1RN
MEFV
PRF1
UNC13D
STX11
STXBP2
NLRP7
CD40LG
AICDA
SERPING1
DOCK8

Primary immunodeficiencies (continued)

- o Hyper-IgE syndrome
- o Hyper-IgD syndrome, HIDS
- o Inflammatory Bowel Disease, IBD
- o Inflammatory Bowel Disease, IBD
- o JPM syndrome, Candle syndrome, Nakajo syndrome
- o Lymphoproliferative syndrome, X-linked, type 1, XLP1
- o Lymphoproliferative syndrome, X-linked, type 2, XLP2
- o Mevalonate kinase deficiency, MKD
- o Muckle-Wells syndrome
- o PAPA syndrome
- o Psoriasis, generalized pustular
- o Severe combined immunodeficiency, X-gebonden SCID, Common γ chain deficientie
- o Severe combined immunodeficiency, SCID
- o Severe combined immunodeficiency, SCID
- o Severe combined immunodeficiency, SCID
- o Severe combined immunodeficiency, SCID
- o Severe combined immunodeficiency, SCID
- o Severe combined immunodeficiency, SCID
- o TNFR associated periodic fever syndrome, TRAPS
- o Wiskott-Aldrich syndrome

STAT3
MVK
IL10RA
IL10RB
PSMB8
SH2D1A
XIAP
MVK
NLRP3
PSTPIP1
IL36RN
IL2RG

ZAP70
CD3G
CD3D
CD3E
RAG1
RAG2
TNFRSF1A
WAS

Renal disease

- o Atypical hemolytic uremic syndrome 1, AHUS1
- o Atypical hemolytic uremic syndrome 2, AHUS2
- o Atypical hemolytic uremic syndrome 3, AHUS3
- o Branchiootorenal syndrome 1, BOR1
- o Branchiootorenal syndrome 2, BOR2
- o Branchiootorenal syndrome 3, BOS3
- o Branchiootorenal syndrome, BOS1
- o Familial vesicoureteral reflux, VUR1
- o Familial vesicoureteral reflux, VUR2
- o Focal segmental glomerulosclerosis 1, FSGS1
- o Focal segmental glomerulosclerosis 2, FSGS2
- o Focal segmental glomerulosclerosis 3, FSGS3

CFH
CD46
CFI
EYA1
SIX5
SIX1
EYA1
PAX2
ROBO2
ACTN4
TRPC6
CD2AP

o Focal segmental glomerulosclerosis 5, FSGS5	INF2	Other diseases (continued)	
o Gitelman syndrome	SLC12A3	o Diarrhea 2, met microvillus atrophy(DIAR2)	MYO5B
o Hirschsprung disease 3, susceptibility to, HSCR3	GDNF	o Fragile-X syndrome, FRAXA included	FMR1
o Hypoparathyroidism, sensorineural deafness, and renal dysplasia	GATA3	o Hemochromatosis, HFE	HFE
o Interstitial lung disease, nephrotic syndrome	ITGA3	o Hemophilia A, HEMA	F8
o Joubert syndrome type 3, JBTS3	AHI1	o Hypothyroidism (CHTE)	IGSF1
o Joubert syndrome type 4, JBTS4	NPHP1	o Infertility, DSD, POF7	AR
o Joubert syndrome type 12, JBTS12	KIF7	o Kennedy disease; X-linked type 1, SBMA, SMAX1	NR5A1
o Nephronophthisis -1	NPHP1	o Lesch-Nyhan syndrome, LNS	HPRT1
o Nephronophthisis -3	NPHP1	o Lung diseases (SMDP3)	ABCA3
o (Nephrogenic) diabetes insipidus	NPHP2	o Persistent Mullerian duct syndrome, PMDS, type 1	AMH
o (Nephrogenic) central diabetes insipidus	AQP2	o Persistent Mullerian duct syndrome, PMDS, type 2	AMHR2
o (Nephrogenic) X- linked diabetes insipidus	AVP	o Prader-Willi syndrome (methylation sensitive MLPA)	[15q11-q13]
o Nephrotic syndrome, congenital Finnish type, NPHS1	AVPR2	o Premature ovarian failure, POF	FMR1
o Nephrotic syndrome, steroid resistant, NPHS2	NPHS1	o Pseudohypoparathyroidism type 1a, PHP1a	GNAS
o Nephrotic syndrome type 3, early onset, NPHS3	NPHS2	o Rendu Osler Weber syndrome, HHT1	ENG
o Nephrotic syndrome met diffuse mesangial sclerosis, NPHS4	PLCE1	o Rendu Osler Weber syndrome, HHT2	ACVRL1
o Pierson syndrome, congenital	WT1	o Rendu Osler Weber syndrome, JPHT	SMAD4
o Renal coloboma syndrome	LAMB2	o Rett syndrome, RTT	MECP2
o Renal adysplasia	PAX2	o Rett syndrome, atypical	CDKL5
o Renal adysplasia	RET	o Rett syndrome, congenital variant	FOXG1
o Renal cysts and diabetes syndrome	UPK3A	o Spinal muscular atrophy type 1 t/m 4, SMA	SMN1/SMN2
	HNF1B	o Susceptibility to Hirschsprung Disease 3, HSCR3	GDNF
Other diseases		o Thrombocytopenia	THPO
o 22q11 microdeletion syndrome, VCFS	[22q11]	o Thrombocytopenia, congenital amegakaryocytic, CAMT	MPL
o Angelman syndrome, AS (methyleringsgevoelige MLPA)	[15q11-q13]	o Uniparental disomy, chromosome:.....	[MARK]
o Angelman syndrome, AS	UBE3A	o Vesicoureteral reflux / renal hypoplasie, VUR1	PAX2
o Azoö/oligozoöspermia [AZF]Adrenal hypoplasia, X-linked, AHC	NR0B1	o X-chromosome inactivation	[AR region]

Confidentiality

The confidentiality of data is guaranteed and secured by the UMC Utrecht guidelines .

Use patient material

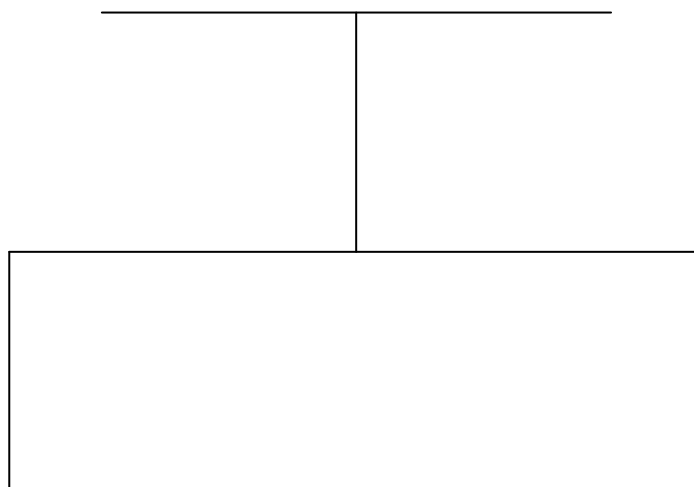
For developing new and improved techniques, the UMC Utrecht uses leftover patient material. It is also used for controls, validation and research in line with the originally diagnostic request.

The referring physician is required to inform the patients about this policy.



The section genome diagnostics has been certified with NEN-EN-ISO 15189:2007 by the Accreditation Council. The scope of accreditation number M001 can be seen on www.rva.nl.

FAMILY PEDIGREE (→ to be investigated; ■/● affected, please state name and date of birth for all relatives previously tested)



Number in family tree	Name	Date of birth

Patient clinical details:

Visit and Correspondence address: Department of Medical Genetics KC.04.084.2, University Medische Genetica, Medical Center Utrecht, Lundlaan 6, 3584 EA Utrecht; PO Box 85090, 3508 AB Utrecht; The Netherlands <http://www.umcutrecht.nl/Genome-Diagnostics>

The UMC Utrecht comprises the University Hospital Utrecht (AZU), the Medical Faculty and the Wilhelmina Children's Hospital (WKZ)

Section Genome Diagnostics
Medical Center Utrecht**PATIENT INFORMATION****Use of patient material**

Please handover this information to the patient

You have donated tissue (e.g. blood, urine, skin biopsy, buccal tissue) for DNA testing. During testing we usually do not use all of the material and are left with a small amount, the leftover material. Our laboratory stores this leftover material indefinitely.

This leftover material is often useful for development of new and improved techniques. It can additionally be used for controls and validations.

The laboratory may also use the leftover material for further testing/research in line with the original diagnostic request. If this leads to relevant clinical findings, your physician will be informed.

What do you have to do?

If you do not have objections about the use of your leftover material, you don't have to do anything.

If you do have objections you can discuss this with your physician. In this case we will not use your leftover material and your physician should record this on the request form.

We hope you are fully informed.

If you have additional questions you can discuss this with your physician.

The text and rules of conduct are available on the website of the Federation of Medical scientific societies (FMWV): www.federa.org.



The section genome diagnostics has been certified with NEN-EN-ISO 15189:2007 by the Accreditation Council. The scope of accreditation number M001 can be seen on www.rva.nl.