Department of Medical Genetics **REQUEST FORM**

Section Genome Diagnostic
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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
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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
Counselee does not grant permission for anonymous use of leftover material (see last page: use of patient material).
O 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)
O Blood (2 x 10 ml EDTA, 2 x 3 ml for neonates)
O Chorionic villi
O Amniotic fluid
O Blood for RNA isolation (2 x 2,5 ml PAXgene blood tubes) (only after consultation)
O DNA number(s) ……
O Tissue type ……
number of sample(s) ……

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

FAMILY HISTORY
O Mutation unknown
O Mutation known
(relation to index patient given in family tree on reverse of form)
Gene: .....
Mutation: .....
Family number: .....
Reference: ……

To be completed by laboratory

U-nummer

Datum::

ETIKETTEN

REGISTRATIE

Indicatie:

Gericht / Volledig

Paraaf: Ontvangstdatum:

Gene panel analysis

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 polymorfic 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)
- ☐ Loeys-Dietz (LDS)

ACTA2, CBS, COL3A1, COL5A1, COL5A2, ELN, FBN1, FBN2, MYH11, MYLK, SLC2A10, SLC8A1, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2

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, CHRNB2, ELP4, GRIN2A, KCNT1, LGI1, SRPX2, SYN1

Deletion test ☐CHRNA4 ☐CHRNB2

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, FOXG1, 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 ☐FOXG1 ☐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/Immunedisregulation (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

TESTEN RvA M 001

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**.

Gene analysis (individual genes)

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 AI4	DLX3
o Cantú syndrome	ABCC9
o Cleidocranial dysplasia, CCD	RUNX2
o Currarino syndrome, TRIAD	MNX1
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	CHRNB2
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, ULD	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, familiar 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

Gene analysis (individual genes)

Metabolic disease (continued)

o Fabry disease, alpha-galactosidase A deficiency	GLA
o Glycerol kinase deficiency, GKD	GK
o Glycine encephalopathy / nonketotic hyperglycinemia	AMT
o Glycine encephalopathy / nonketotic hyperglycinemia	GCSH
o Glycine encephalopathy / nonketotic hyperglycinemia	GLDC
o Hartnup disorder	SLC6A19
o Hyperinsulinemic hypoglycemia type 7, HHF7	SLC16A1
o Metachromatic leukodystrophy, MLD	ARSA
o Methylmalonic aciduria type cblA	MMAA
o Phenylketonuria type 1, PKU	PAH
o Phenylketonuria, hyperphenylalaninemia, BH4-deficient, PTPS	PTS
o Pompe disease, Glycogen storage disease II, GSD2	GAA
o Pyruvate kinase deficiency, PK	PKLR
o Serine biosynthesis defect, PHGDH deficiency	PHGDH
o Serine biosynthesis defect, PSPH deficiency	PSPH
o Serine biosynthesis defect, PSAT1 deficiency	PSAT1
o Tyrosinemia, type I	FAH
o Wilson disease, WD	ATP7B

Neurological disease

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

Neuromuscular disease

o Ehlers-Danlos syndrome (musculocontractural)	CHST14
o Minicore myopathy/ Central core disease	RYR1
o Muscular dystrophy, Emery-Dreifuss type 6, EDMD6	FHL1
o Muscular dystrophy, Limb-Girdle type 2G, LGMD2G	TCAP
o Myotonic dystrophy type 1, DM1	DMPK
o Myotonic dystrophy type 2, DM2	CNBP
o Myofibrillar myopathy type 1. MFM1	DES
o Myofibrillar myopathy type 2, MFM2	CRYAB
o Nemaline myopathy type 1, NEM1	TPM3
o Nemaline myopathy type 3, NEM3	ACTA1
o Nemaline myopathy type 4, NEM4	TPM2
o Nemaline myopathy type 5, NEM5	TNNT1
o Nemaline myopathy type 6, NEM6	KBTBD13
o Nemaline myopathy type 7, NEM7	CFL2
o Spinal Muscular Atrophy (SMA type 1 - 4)	SMN1

Obesity

o Alstrom syndrome	ALMS1
o Cohen syndrome	VPS13B
o Leptin deficiency	LEP
o Leptin receptor deficiency	LEPR
o Obesity with impaired prohormone processing	PCSK1
o Proopiomelanocortin deficiency	POMC
o Obesity, autosomal dominant	MC4R

Primary immunodeficiencies

o Acne inversa, familiar type 1	NCSTN
o Acne inversa, familiar type 2	PSENEN
o Agammaglobulinemia, X-linked, XLA	BTK
o Autoimmune lymphoproliferative syndrome, ALPS type 1a	FAS
o Autoimmune lymphoproliferative syndrome, ALPS type 1b	FASL
o Autoimmune lymphoproliferative syndrome, ALPS type 2a	CASP10
o Autoimmune polyendocrinopathy syndrome, type I, APS1	AIRE
o Blau syndrome	NOD2
o Candidiasis, familiar type 2	CARD9
o Candidiasis, familiar type 5	IL17RA
o Candidiasis, familiar type 6	IL17F
o Candidiasis, familiar type 7	STAT1
o CINCA syndrome	NLRP3
o Cold-induced autoinflammatory syndrome FCAS1	NLRP3
o Cold-induced autoinflammatory syndrome FCAS2	NLRP12
o Cold-induced autoinflammatory syndrome FCAS3	PLCG2
o DIRA syndrome	IL1RN
o Familial Mediterranean fever, FMF	MEFV
o Hemophagocytic lymphohistiocytosis, HLH type 2	PRF1
o Hemophagocytic lymphohistiocytosis, HLH type 3	UNC13D
o Hemophagocytic lymphohistiocytosis, HLH type 4	STX11
o Hemophagocytic lymphohistiocytosis, HLH type 5	STXBP2
o Hydatidiform mole, recurrent type 1	NLRP7
o Hyper-IgM syndrome, CD40 ligand deficiency	CD40LG
o Hyper-IgM syndrome, AID deficiency	AICDA
o Hereditaire Angioderma type 1	SERPING1
o Hyper-IgE syndrome	DOCK8

Primary immunodeficiencies (continued)

o Hyper-IgE syndrome	STAT3
o Hyper-IgD syndrome, HIDS	MVK
o Inflammatory Bowel Disease, IBD	IL10RA
o Inflammatory Bowel Disease, IBD	IL10RB
o JPM syndrome, Candle syndrome, Nakajo syndrome	PSMB8
o Lymphoproliferative syndrome, X-linked, type 1, XLP1	SH2D1A
o Lymphoproliferative syndrome, X-linked, type 2, XLP2	XIAP
o Mevalonate kinase deficiency, MKD	MVK
o Muckle-Wells syndrome	NLRP3
o PAPA syndrome	PSTPIP1
o Psoriasis, generalized pustular	IL36RN
o Severe combined immunodeficiency, X-gebonden SCID, Common γ chain deficiëntie	IL2RG
o Severe combined immunodeficiency, SCID	ZAP70
o Severe combined immunodeficiency, SCID	CD3G
o Severe combined immunodeficiency, SCID	CD3D
o Severe combined immunodeficiency, SCID	CD3E
o Severe combined immunodeficiency, SCID	RAG1
o Severe combined immunodeficiency, SCID	RAG2
o TNFR associated periodic fever syndrome, TRAPS	TNFRSF1A
o Wiskott-Aldrich syndrome	WAS

Renal disease

o Atypical hemolytic uremic syndrome 1, AHUS1	CFH
o Atypical hemolytic uremic syndrome 2, AHUS2	CD46
o Atypical hemolytic uremic syndrome 3, AHUS3	CFI
o Branchiootorenal syndrome 1, BOR1	EYA1
o Branchiootorenal syndrome 2, BOR2	SIX5
o Branchiootorenal syndrome 3, BOS3	SIX1
o Branchiootic syndrome, BOS1	EYA1
o Familiar vesicoureteral reflux, VUR1	PAX2
o Familiar vesicoureteral reflux, VUR2	ROBO2
o Focal segmental glomerulosclerosis 1, FSGS1	ACTN4
o Focal segmental glomerulosclerosis 2, FSGS2	TRPC6
o Focal segmental glomerulosclerosis 3, FSGS3	CD2AP

Gene analysis (individual genes)

o Focal segmental glomerulosclerosis 5, FSGS5	INF2
o Gitelman syndrome	SLC12A3
o Hirschsprung disease 3, susceptibilty to, HSCR3	GDNF
o Hypoparathyroidism, sensorineural deafness, and renal dysplasia	GATA3
o Interstitial lung disease, nephrotic syndrome	ITGA3
o Joubert syndrome type 3, JBTS3	AHI1
o Joubert syndrome type 4, JBTS4	NPHP1
o Joubert syndrome type 12, JBTS12	KIF7
o Nephronophthisis -1	NPHP1
o Nephronophthisis -3	NPHP3
o (Nephrogenic) diabetes insipidus	AQP2
o (Nephrogenic) central diabetes insipidus	AVP
o (Nephrogenic) X- linked diabetes insipidus	AVPR2
o Nephrotic syndrome, congenital Finnish type, NPHS1	NPHS1
o Nephrotic syndrome, steroid resistant, NPHS2	NPHS2
o Nephrotic syndrome type 3, early onset, NPHS3	PLCE1
o Nephrotic syndrome met diffuse mesangial sclerosis, NPHS4	WT1
o Pierson syndrome, congenital	LAMB2
o Renal coloboma syndrome	PAX2
o Renal adysplasia	RET
o Renal adysplasie	UPK3A
o Renal cysts and diabetes syndrome	HNF1B

Other diseases

o 22q11 microdeletion syndrome, VCFS	[22q11]
o Angelman syndrome, AS (methyleringsgevoelige MLPA)	[15q11-q13]
o Angelman syndrome, AS	UBE3A
o Azoö/oligozoöspermia [AZF]Adrenal hypoplasia, X-linked, AHC	NR0B1

Other diseases (continued)

o Diarrhea 2, met microvillus atrophy(DIAR2)	MYO5B
o Fragile-X syndrome, FRAXA included	FMR1
o Hemochromatosis, HFE	HFE
o Hemophilia A, HEMA	F8
o Hypothyroidism (CHTE)	IGSF1
o Infertility, DSD, POF7	NR5A1
o Kennedy disease; X-linked type 1, SBMA, SMAX1	AR
o Lesch-Nyhan syndrome, LNS	HPRT1
o Lung diseases (SMDP3)	ABCA3
o Persistent Mullerian duct syndrome, PMDS, type 1	AMH
o Persistent Mullerian duct syndrome, PMDS, type 2	AMHR2
o Prader-Willi syndrome (methylation sensitive MLPA)	[15q11-q13]
o Premature ovarian failure, POF	FMR1
o Pseudohypoparathyroidism type 1a, PHP1a	GNAS
o Rendu Osler Weber syndrome, HHT1	ENG
o Rendu Osler Weber syndrome, HHT2	ACVRL1
o Rendu Osler Weber syndrome, JPHT	SMAD4
o Rett syndrome, RTT	MECP2
o Rett syndrome, atypical	CDKL5
o Rett syndrome, congenital variant	FOXG1
o Spinal muscular atrophy type 1 t/m 4, SMA	SMN1/SMN2
o Susceptibilty to Hirschsprung Disease 3, HSCR3	GDNF
o Thrombocythemia	THPO
o Thrombocytopenia, congenital amegakaryocytic, CAMT	MPL
o Uniparental disomy, chromosome:……	[MARK]
o Vesicoureteral reflux / renal hypoplasie, VUR1	PAX2
o X-chromosome inactivation	[AR region]

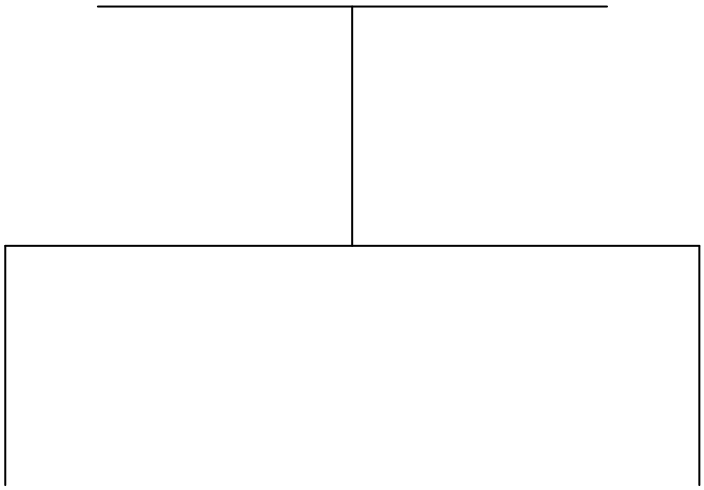
Confidentiality
The confidentiality of data is guaranteed and secured by the UMC Utrecht guidelines .

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.

TESTEN RvA M 001

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

Section Genome Diagnostics
Medical Center Utrecht

UMC 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.

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