

Department of Medical Genetics**APPLICATION FORM DNA-TESTING****Genome Diagnostics Section**

DNA-diagnostics Laboratory
 University Medical Center Utrecht
 Centrale Balie CDL, G.03.3.30
 Heidelberglaan 100
 3584 CX Utrecht
 The Netherlands

**Administration**

Tel +31 (0)88-7554090
 Fax +31 (0)88-7555034
 E-mail genoomdiagnostiek@umcutrecht.nl

Personal data

name
 date of birth
 gender
 address
 country

Referring physician

Name:

Hospital:

Address:

City:

Date:

Your ref:

Department:

Telephone:

Zip code:

INDICATION

Mark on the table (see reverse)

State relevant clinical information and/or draw pedigree (on reverse)

MATERIAL

Tubes of blood (or DNA samples) please label clearly with name, gender and date of birth
Urgent and prenatal requests: please phone first to discuss (tel. +31 (0)88-7554090)

- Blood (2 x 10 ml EDTA, 2 x 3 ml for little children)
- Chorionic Villi
- Amniotic cells
- Blood for RNA isolation (2 x 2,5 ml PAXgene blood tubes) ([only after consultation](#))
- DNA number(s)
- Tissue type
- number sample(s)

PURPOSE

- Confirmation of clinical diagnosis / exclusion of a diagnosis
- Carriership (with respect to a 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)
 Mutation:
- Family number:
- Reference:

In te vullen door LABORATORIUM**ETIKETTEN****REGISTRATIE**

U-nummer

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Ontvangstdatum:

Wijze: Gericht / Volledig

Paraaf:

Datum:

INDICATION	GENE	INDICATION	GENE
o 22q11 microdeletie-syndroom, VCFS	[22q11]	o Myotonic dystrophy type 1, DM1	DMPK
o Acromegaly deletion; pituitary adenoma predisposition	AIP	o Myotonic dystrophy type 2, DM2	ZNF9
o Adrenal hypoplasia, X-linked, AHC	NROB1	o Nephronophthisis	NPHP1
o Albright hereditary osteodystrophy, AHO	GNAS	o Oligodontia/hypodontia	AXIN2
o Amyloidosis I and VII; transthyretin amyloidosis	TTR	o Oligodontia/hypodontia	IRF6
o Amyotrophic lateral sclerosis type 1, ALS1	SOD1	o Oligodontia/hypodontia	SUMO1
o Amyotrophic lateral sclerosis type 6, ALS6	FUS	o Oligodontia/hypodontia	TBX22
o Amyotrophic lateral sclerosis type 8, ALS8	VAPB	o Oligodontia-colo-rectal cancer syndrome	AXIN2
o Amyotrophic lateral sclerosis type 9, ALS9	ANG	o PAPA syndrome	PSTPIP1
o Amyotrophic lateral sclerosis type 10, ALS10	TARDBP	o Papillary renal cell carcinoma, PRC	MET
o Angelman syndrome	[AS]	o Phenylketonuria type 1. phenylalanin hydroxylase deficiency, PKU	PAH
o Arrhythmogenic right ventricular dysplasia, ARVD/C1	TGFB3	o Phenylketonuria type 3. 6-pyruvoyl tetrahydro pterin synthase deficiency, PTPS	PTS
o Arrhythmogenic right ventricular dysplasia, ARVD/C5	TMEM43	o Pheochromocytoma/paraganglioma, hereditary	SDHB
o Arrhythmogenic right ventricular dysplasia, ARVD/C 8	DSP	o Pheochromocytoma/paraganglioma, hereditary	SDHC
o Arrhythmogenic right ventricular dysplasia, ARVD/C 9	PKP2	o Pheochromocytoma/paraganglioma, hereditary	SDHD
o Arrhythmogenic right ventricular dysplasia, ARVD/C 10	DSG	o Polyneuropathy, amyloïde	TTR
o Arrhythmogenic right ventricular dysplasia, ARVD/C 11	DSC2	o Pompe disease, GSD2	GAA
o Arrhythmogenic right ventricular dysplasia, ARVD/C 12	JUP	o Prader-Willi syndrome, PWS	[PWS]
o Azoöspermia; severe oligozoöspermia, AZF	[AZF]	o Premature ovarian failure, POF	FMR1
o Blau syndrome	NOD2	o Pseudohypoparathyroidism type 1a, PHP1a	GNAS
o Breast cancer, hereditary, BRCA1	BRCA1	o Pyruvate kinase deficiency, PK	PKLR
o Breast cancer, hereditary, BRCA2	BRCA2	o Renal coloboma syndrome	PAX2
o Cataract and dilating cardiomyopathy, CRYAB	CRYAB	o Renal adysplasia	UPK3A
o Cholestasis, familial intrahepatic 1, P	ATP8B1	o Rendu, Osler and Weber syndrome, HHT1	ENG
o Cholestasis, familial intrahepatic 2, PFIC2	ABCB11	o Rendu, Osler and Weber syndrome, HHT2	ACVRL1
o CINCA syndrome	NLRP3	o Rendu, Osler and Weber syndrome, JPHT	SMAD4
o Currarino triad, TRIAD	HLXB9	o Rett syndrome, RTT	MECP2
o DIRA syndrome	IL1RN	o Rett syndrome, atypical	CDKL5
o Fabry disease; alpha-galactosidase A deficiency, FABRY	GLA	o Schizencephaly	EMX2
o Fallot, Tetralogy of, TOF	NKX2-5	o Spinocerebellar ataxia, autosomal dominant, SCA1	ATXN1
o Familial cold auto inflammatory syndrome 1, FCAS1	NLRP3	o Spinocerebellar ataxia, autosomal dominant, SCA2	ATXN2
o Familial mediterranean fever, FMF	MEFV	o Spinocerebellar ataxia, autosomal dominant, SCA3	ATXN3
o Familial platelet disorder, with associated myleoid malignancy	CEBPA	o Spinocerebellar ataxia, autosomal dominant, SCA6	CACNA1A
o Fragile-X syndrome, FRAXA	FMR1	o Spinocerebellar ataxia, autosomal dominant, SCA7	ATXN7
o Hyperglycerolemia	GK	o Spinocerebellar ataxia, autosomal dominant, SCA12	PP2R2B
o Hemochromatosis HFE	HFE	o Spinocerebellar ataxia, autosomal dominant, SCA13	KCNC3
o Hemophilia A, HEMA	F8	o Spinocerebellar ataxia, autosomal dominant, SCA14	PRKCG
o Hyper-IgD syndrome, HIDS	MVK	o Spinocerebellar ataxia, autosomal dominant, SCA17	TBP
o Hyperparathyroidism, familial primary, HRPT1	MEN1	o Sporadic medullary thyroid carcinoma, MTC	RET
o Hypodontia, familial, HYD1	MSX1	o Tumor necrosis factor receptor-associated periodic fever syndrome, TRAPS	TNFRSF1A
o Hypodontia, familial, HYD3	PAX9	o Tyrosinemia type 1, HT1	FAH
o Joubert syndrome type 3, JBTS3	AHI	o Uniparental disomy, chromosome:.....	[MARK]
o Joubert syndrome type 4, JBTS4	NPHP1	o Vesicoureteral reflux1; VUR1.....	PAX2
o Kennedy disease; SBMA, X-linked type 1, SBMA, SMAX1	AR	o Von Hippel-Lindau disease	VHL
o Lesch-Nyhan syndrome, LNS	HPRT1	o Van der Woude syndrome, VWS	IRF6
o Lynch syndrome, HNPCC2	MLH1	o Wilson Disease, WD	ATP7B
o Lynch syndrome, HNPCC1	MSH2	o X-chromosome inactivaton	AR
o Lynch syndrome, HNPCC5	MSH6	o Zygosity test	[X2Y]
o Medium-chain acyl-coA dehydrogenase deficiency, MCAD	ACADM	o Other.....	
o Metachromatic leukodystrophy, MLD	ARSA		
o Mevalonate kinase deficiency, MKD	MVK		
o Microsatellite instability test	[MSI]		
o Muckle-Wells syndrome	NLRP3		
o Multiple endocrine neoplasia type 1A, MEN1	MEN1		
o Multiple endocrine neoplasia type 2A, MEN2A	RET		
o Multiple endocrine neoplasia type 2B, MEN2B	RET		
o Multiple endocrine neoplasia type 4, MEN4	CDKN1B		

INDICATION**GENE****Epilepsy**

o Alpers-Huttenlocher syndrome	POLG
o Autosomal dominant lateral temporal lobe epilepsy, ADLTE, LGI1)	LGI1
o Benign familial neonatal convulsions, BFNC	KCNQ2
o Benign familial neonatal convulsions, BFNC	KCNQ3
o Benign familial neonatal-infantile seizures, BFNIS	SCN2A
o Female restricted epilepsy with mental retardation, EFMR	PCDH19
o Generalized epilepsy with febrile seizures plus, GEFS+	SCN1A
o Generalized epilepsy with febrile seizures plus, GEFS+	SCN1B
o Generalized epilepsy with febrile seizures plus, GEFS+	SCN2A
o Generalized epilepsy with febrile seizures plus, GEFS+	GABRG2
o Epileptic encephalopathy, early infantile 2	CDKL5
o Nocturnal frontal lobe epilepsy, type 1, ADNFLE1	CHRNA4
o Nocturnal frontal lobe epilepsy, type 3, ADNFLE3	CHRNA2
o Progressive myoclonic epilepsy type 1 (EPM1): CSTB gene	CSTB
o Progressive myoclonic epilepsy type 2A / lafora body disease, EPM2A	EPM2A
o Progressive myoclonic epilepsy type 2B / lafora body disease, EPM2B	NHLRC1
o Progressive myoclonic epilepsy type 3, EPM3	KCTD7
o Pyridoxin-dependent epilepsy, PDE	ALDH7A1
o Pyridoxin-dependent epilepsy, PDE	PNPO
o Severe myoclonic epilepsy of infancy, SMEI, SCN1A	SCN1A
o Unverricht-Lundborg disease, EPM1	CSTB

Primary Immunodeficiencies (only after consultation)

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 Hemophagocytic lymphohistiocytosis, familial; HLH type 2	PRF1
o Hemophagocytic lymphohistiocytosis, familial; HLH type 3	UNC13D
o Hemophagocytic lymphohistiocytosis, familial; HLH type 4	STX11
o Immunodeficiency with hyper-IgM, CD40 ligand deficiency	CD40LG
o Immunodeficiency with hyper-IgM, AID deficiency	AICDA
o ICOS deficiency	ICOS
o Severe combined immunodeficiency; X-linked SCID,	IL2RG
o Severe combined immunodeficiency; SCID	JAK3
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 Wiskott-Aldrich syndrome	WAS
o Lymphoproliferative syndrome, type 1, XLP1	SH2D1A
o Lymphoproliferative syndrome, type 2, XLP2	XIAP

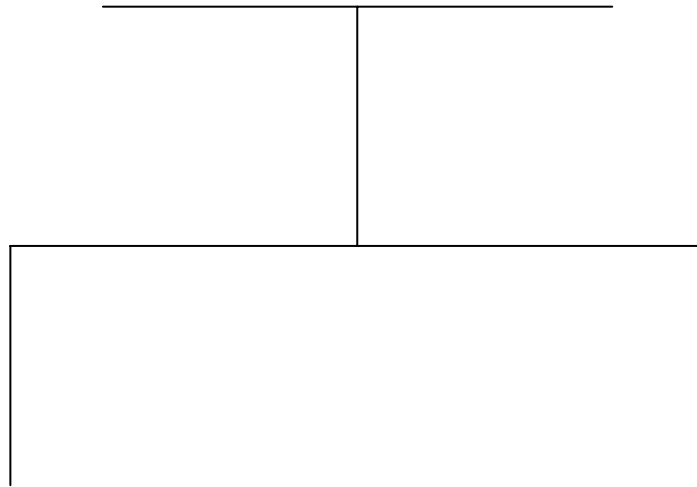
For disorders and gene tests which are not mentioned: The application forms for the Clinical Genetic Centres in the Netherlands are available on the LOD-website (www.dnadiagnostiek.nl). Each centre has its own specialisation regarding the diagnostics offered. The Internet site gives a complete and up to date overview.



The genome diagnostics section 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.

The tests covered by Dutch insurance companies are updated regularly. You can download and print the most recent version of this application form from our website <http://www.umcutrecht.nl/Genome-Diagnostics/>.

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



Number in family tree	Name	Date of birth	D-nummer

Relevant clinical information on person requesting the DNA test / Remarks: