



DEPARTMENT OF CLINICAL GENETICS
SECTION GENOME DIAGNOSTICS (GD)

REQUISITION FORM FOR MOLECULAR GENETIC TESTING

The section GD is NEN-EN-ISO
15189:2022 accredited by the
Dutch Accreditation Council. The
scope for accreditation number
M007 can be found at www.rva.nl.



Please fully complete the form (one form per person).

Surname and Initials*

Name spouse

Street name and number*

Postal code and city* Country*

Date of birth* (yyyy/mm/dd)

Sex*

* REQUIRED FIELDS

Patient information / Fill out completely

Postal address

LUMC, Building 2
KG, Genome diagnostics S-06-P
Freepost 10392, 2300 WB Leiden
The Netherlands

Visiting address/ Courier service: Einthovenweg 20,
2333 ZC Leiden

Administration:

Tel: +3171-5269800
Email: genoomdiagnostiek@lumc.nl
Website: www.LUMC.nl/klingen

PROCEDURE:

Always consult us prior to sending material other than blood or DNA. Tel: +31715269800.
All materials must be clearly labelled with number, name and date of birth of the patient.

MATERIAL:

- DNA TESTING: 8-10 ml EDTA blood (neonates ≥ 2.5 ml), DNA (at least 15 μ g), tissue, chorionic villi (20 mg) or amniotic fluid (15 ml).
Please note for FSHD & Hemophilia 2 tubes EDTA blood.
- RNA TESTING: Use the "RNA ANALYSIS form".

TRANSPORT:

EDTA blood and DNA can be sent at room temperature by post to the address above. Use an overnight courier for priority samples and cooled material.

PATIENT INFORMATION:

Please give to the patient, this can be found at <https://www.lumc.nl/en/afdelingen/clinical-genetics/request-forms/>
For diagnostic turnaround times, our current criteria for diagnostic requests and opening hours, see our website.

When requesting this genetic test, we assume that the risk of incidental findings was discussed with the patient.

Objection to other use of remaining material: **yes** **no**

Due to incomplete applications there is a possibility of delay

REFERRING PHYSICIAN :	Telephone :
Hospital/Institution :	Department :
Address :	Your ref. no. :
Postal code / City :	Email :
Date of collection :	

REASON FOR REFERRAL

- | | |
|--|--|
| ☐ carrier testing (for recessive diseases only) | ☐ prenatal testing (only after consultation) |
| ☐ confirmation / exclusion of clinical diagnosis | ☐ request for interpretation of variant in index patient |
| ☐ predictive / presymptomatic testing | ☐ Only storage, reason: |
| ☐ testing for family members | |

GENE(S) / TEST:

(see next pages for overview)

Did you previously send us material from this patient, a family member or spouse?

☐ NO ☐ YES (patient) ☐ YES (family members, fill in table)

Known mutation: yes:

LDGA Family number (F-No.):

CLINICAL INFORMATION and/or **PEDIGREE** (draw pedigree after print or add separately, indicate index with arrow):

Information of tested family members:

No. In pedigree	Name (full)	Date of birth	Sex	Relation to current patient

TO BE FILLED OUT BY PATIENT SECRETARY:

Datum ontvangst:

Paraaf ontvangst:

Materiaal en aantal: Bloed / DNA / Vlokken / Vruchtwater/Weefsel

Familienummer:

☐ Alleen formulier

Gene panels

See next pages for request of individual genes

- Basal cell Carcinoma
- o Breast and ovarium cancer panel
- o Cerebral angiopathies / adult-onset leukoencephalopathies
(including CADASIL)
- o Coffin-Siris / Nicolaidis-Baraitser syndrome
- o Colorectal carcinoma
- o Episodic Ataxia
- o FAMMM (Familial Atypical Multiple Mole-Melanoma)
- o Familial pancreatic carcinoma
- o Short stature, basic gene panel
- o Hereditary Multiple Osteochondromas
- o LYNCH syndrome
- o Lipodystrophy
- o Migraine, familial hemiplegic
- o MODY (Maturity Onset Diabetes of the Young)
- o Muscular dystrophies / myopathies
- o Paragangliomas and/or pheochromocytomas
- o Polyglutamin repeat disorders
- o Polyposis coli, adenomatous*
- o Polycystic kidney disease
- o Skeletal Muscle Channelopathies

Alias

- BCC panel**
- HBOC panel**
- CHA panel**
- CSS panel**
- CRC panel**
- EA panel**
- Melanoma panel**
- PACA panel**
- Growth panel**
- HMO panel**
- LYNCH panel**
- LIPO panel**
- FHM panel**
- Diabetes panel/ MODYScan**
- Muscle panel/ MuscleScan**
- PGL panel**
- PolyQ**
- Polyp panel**
- PKD panel**
- Channelopathies**

For an overview of all genes in the gene panels see: <https://www.lumc.nl/over-het-lumc/afdelingen/klinische-genetica/genpanels/>

NB. NGS is performed by GenomeScan B.V.

Genome analysis

- o Mental retardation or developmental delay, with or without multiple congenital defects
- o Microdeletion syndrome (specify)
- o Growth disorders
- o Carrier detection as a result of CNV finding

Test

- o CNV analysis (genome wide)
- o CNV analysis (genome wide)
- o CNV analysis (genome wide)
- o CNV analysis (genome wide)

Disorder/Referral	Type	Gene/Test
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Blood diseases

- | | | |
|---|--------|-------|
| o Hemochromatosis | Type 1 | o HFE |
| o Hemoglobinopathies / Thalassemia | | |
| Please use "Requisition form Hemoglobinopathy analysis" | | |
| o Hemophilia (<i>Please send in 2 tubes EDTA blood</i>) | Type A | o F8 |
| | Type B | o F9 |

Cancer genetics

*Requests only by a consultant clinical geneticist

- | | | |
|--|-----|--|
| o Breast- and ovarian cancer, hereditary * | | ATM
BARD1
BRCA1
BRCA2
BRIP1
CHEK2
PALB2
PTEN |
| o Clear cell meningioma/ Familial Multiple Meningioma* | CCM | o RAD51C
o RAD51D
o SMARCE1
o SMARCB1 |
| o FAMMM (Familial Atypical Multiple Mole-Melanoma)* | | o CDKN2A
o CDK4
o POT1
o BAP1
o MITF
o SDHA |
| o Gastrointestinal Stromal Tumors
(GIST, Carney-Stratakis syndrome) | | |
| o Hyperparathyroidism-jaw tumor syndrome (HPT-JT/HRPT2) | | o CDC73 |
| o Lynch syndrome (HNPCC)* | | o MLH1 |
| | o | MSH2 (incl. EPCAM) |
| | | o MSH6 |
| | | o PMS2 |
| o Myeloproliferative diseases (MPDs, somatic mutation) | | o JAK2
(p.Val617Phe) |
| | o | MPN-combi:
JAK2 exon 12 &
exon 14 p.(Val617Phe),
MPL exon 10 and
CALR exon 9 |

Disorder/Referral	Type	Gene/Test
o Parangliomas and/or pheochromocytomas		o MAX
		o SDHA
		o SDHAF2
		o SDHB
		o SDHC
		o SDHD
		o TMEM127
o Polyposis coli, adenomatous*	FAP1	APC (incl. GREM1)
	MAP	o MUTYH
	NAP	o NTHL1
	PPAP	o POLD1
	PPAP	o POLE
	FAP4	o MSH3
o Renal Cell Carcinoma (RCC), hereditary		o SDHB
o Rhabdoid tumor predisposition syndrome (RTPS)*	RTPS1	o SMARCB1
	RTPS2	o SMARCA4
o Small cell carcinoma of the ovary, hypercalcemic type*	SCCOHT	o SMARCA4
	SCCOHT	o SMARCB1
o Schwannomatosis*		o SMARCB1

Channelopathies

o Hyperkalemic periodic paralysis (HYPP)		o SCN4A
o Hypokalemic periodic paralysis (HOKPP)	Type 1	o CACNA1S
	Type 2	o SCN4A
o Myotonia congenita (Thomsen, Becker disease)		o CLCN1
o Myotonia permanens/fluctuans		o SCN4A
o Paramyotonia congenita		o SCN4A

Diabetes

o Hyperproinsulinemia	o INS
o Insulin dependent diabetes	o INS
o MIDD (Maternally Inherited Diabetes and Deafness)	o m.3243A>G tRNALEU/UUR

Disorder/Referral	Type	Gene/Test
o MODY (Maturity Onset Diabetes of the Young)	Type 1	o HNF4A
	Type 2	o GCK
	Type 3	o HNF1A
	Type 4	o PDX1 (IPF1)
	Type 5	o HNF1B
	Type 6	o NEUROD1
	Type 10	o INS
o PNDM (Permanent Neonatal Diabetes Mellitus)		GCK
		o INS
		o KCNJ11
o Persistent hyperinsulinemic hypoglycemia of infancy (PHHI)		o GCK
		o KCNJ11

Growth and skeletal defects

o Achondroplasia	o FGFR3
o Acromesomelic dysplasia Type Maroteaux	o NPR2
o Hereditary Multiple Osteochondromas	o EXT1
	o EXT2
o NPR2- related tall stature	o NPR2
o Hypochondroplasia	o FGFR3
o Langer mesomelic dysplasia (Leri-Weill dyschondrosteosis)	o SHOX
o Multiple epiphyseal dysplasia	o COMP
o Pseudoachondroplastic dysplasia	o COMP
o Short stature (proportionate)	o GH1
	o GHR
	o GHSR
	o IGF1
	o IGF1R
	o IGFALS
	o STAT5B
o Short stature (osteochondritis dissecans)	o ACAN
o Tall stature	o NPR2
o Thanatophoric dysplasia	o FGFR3
o Van Buchem disease	o VBCH

Disorder/Referral	Type	Gene/Test
Immune system		
o Chilblain lupus	Type 1	o TREX1
o Granulomatous disease, chronic, X-linked		o CYBB
o Lymphoproliferative syndrome, X-linked		o XLP
o Mediterranean fever, familial (FMF)		o MEFV
o Wiskott-Aldrich syndrome		o WAS
Metabolic diseases		
o Adrenal hypoplasia, congenital		o NR0B1 (DAX1)
o Cystinuria		o SLC3A1
		o SLC7A9
Muscular dystrophies/ Myopathies		
Slow-channel congenital myasthenic syndrome-4A (CMS4A)	Type 4A	CHRNE
Congenital myasthenic syndrome-5 (CMS5)	Type 5	COLQ
Congenital myasthenic syndrome-9 (CMS9)	Type 9	MUSK
associated with AChR deficiency		
Congenital myasthenic syndrome-10 (CMS10)	Type 10	DOK7
Congenital myasthenic syndrome-11 (CMS11)	Type 11	RAPSN
associated with acetylcholine receptor deficiency		
Congenital myasthenic syndrome-14 (CMS14)	Type 14	ALG2
Congenital myasthenic syndrome-15 (CMS15)	Type 15	ALG14
Duchenne and Becker		DMD MLPA only
		DMD Sequencing only
		DMD MLPA, if
		negative directly followed by
		sequencing
Emery-Dreifuss (X-linked)		EMD
Facioscapulohumeral (FSHD)	Type 1/2	Rearrangement chromosome 4
(Please send in 2 tubes of EDTA blood)		Permissive haplotype analysis
		(4qA/B)
	Type 2	SMCHD1
		LRIF1
		DNMT3B

Disorder/Referral	Type	Gene/Test
o Limb Girdle	Myofibrillar myopathy	o MYOT
	Emery–Dreifuss muscular dystrophy (EDMD)	o LMNA
	Rippling muscle disease	o CAV3
	LGMD D4 / R1	o CAPN3
	LGMD R2	o DYSF
	LGMD R5	o SGCG
	LGMD R3	o SGCA
	LGMD R4	o SGCB
	LGMD R6	o SGCD
	LGMD R7	o TCAP
	LGMD R8	o TRIM32
	LGMD R9	o FKRP
	LGMD R12	o ANO5
o Miyoshi (MMD3)		o ANO5
o Myopathy with extrapyramidal signs		o MICU1
Neurogenetics		
o Aicardi-Goutières syndrome	Type 1	o TREX1
o Alternating Hemiplegia of Childhood	Type 2	o ATP1A3
o CADASIL		o NOTCH3
o CARASIL/ CADASIL	Type 2	o HTRA1
o Cerebral hemorrhage with amyloidosis (HCHWA-D)		o APP
o Dentatorubral-pallidoluysian atrophy (DRPLA)		o ATN1
o Episodic ataxia	Type 2	o CACNA1A
o Huntington disease		o HTT
o Huntington, disease-like 2 (HDL2)		o JPH3
o Hyperekplexia (familial Startle disease)		o GLRA1
		o GLRB
		o SLC6A5
		o ATP1A2
o Migraine, familial hemiplegic (FHM)		o CACNA1A
		o SCN1A
		o SGCE
o Myoclonus dystonia syndrome		

Disorder/Referral	Type	Gene/Test
o Neuronal ceroid lipofuscinosis (NCL) Juvenile	Late infantile	o CLN3
	Late infantile	o TPP1 (CLN2)
	Late infantile	o CLN6
	Late infantile	o CLN8
	Late infantile / adult	o PPT1 (CLN1)
o Paroxysmal torticollis		o CACNA1A
• Polyglutamin repeat disorders		o CACNA1A, TBP, ATXN1, ATXN7, ATXN2, ATXN3 and ATN1
o Retinal vasculopathy with cerebral leukodystrophy (RVCL)		o TREX1
Polycystic kidney disease		
o Autosomal dominant Polycystic kidney disease (ADPKD)	Dominant	o PKD1
	Dominant	o PKD2
o Autosomal dominant Polycystic kidney and liver disease (ADPKD)	Dominant	o GANAB
o Autosomal recessive Polycystic kidney (ARPKD)	Recessive	o PKHD1
o Renal cysts and diabetes syndrome (RCAD)	Dominant	o HNF1B
Syndromes		
o Coffin-Siris syndrome		o ARID1A
		o ARID1B
		o SMARCA4
		o SMARCB1
		o SMARCE1
o Ellis van Creveld syndrome		o EVC
		o EVC2
o Filippi syndrome		o CKAP2L
o Marshall-Smith syndrome		o NFIX
o Nicolaides-Baraitser syndrome		o SMARCA2

Disorder/Referral	Type	Gene/Test
o Peters Plus syndrome		o B3GLCT (B3GALTL)
o Pitt-Hopkins syndrome		o TCF4
o Rubinstein - Taybi syndrome		o CREBBP
		o EP300
o Sotos syndrome		o NSD1
o Sotos-like syndrome		o DNMT3A
		o NFIX
		o SETD2
		o HIST1H1E
o TAR (thrombocytopenia-absent radius) syndrome		o 1q21.1 deletion and RBM8A SNP
o Weaver syndrome		o EZH2
Other		
o Hypocalciuric Hypercalcemia, Familial (FHH)		o CASR
		o GNA11
		o AP2S1
o Keratosis follicularis spinulosa decalvans (KFSD)		o MBTPS2