DEPARTMENT OF CLINICAL GENETICS
SECTION GENOME DIAGNOSTICS (GD)

REQUISITION FORM FOR MOLECULAR GENETIC TESTING

The section GD is NEN-EN-ISO 15189:2012 accredited by the Dutch Accreditation Council. The scope for accreditation number M007 can be found at **www.rva.nl**.

TESTING RvA M 007

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 adress:
LUMC, Building 2
KG, Genome diagnostic S-06-P
Visiting address: Einthovenweg 20,
Reply number 10392
2300 RC Leiden
The Netherlands

Administration:
Tel: 071-5269800
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 µg), 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.

FORM: Please fully complete the form (**one form per person**).

PATIENT INFORMATION: Please give to the patient, this can be found at https://www.lumc.nl/over-het-lumc/afdelingen/klinische-genetica/aanvraagformulieren/

For diagnostic turnaround times, our current criteria for diagnostic requests and opening hours, see our website.

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

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:

O Alleen formulier

REQUISITION FORM FOR MOLECULAR GENETIC TESTING

Gene panels

See next pages for request of individual genes

Alias

☐	Breast and ovarium cancer panel	**HBOC panel**
☐	Cerebral angiopathies / adult-onset leukoencephalopathies (including CADASIL)	**CHA panel**
☐	Coffin-Siris / Nicolaides-Baraitser syndrome	**CSS panel**
☐	Colorectal carcinoma	**CRC panel**
☐	Episodic Ataxia	**EA panel**
☐	FAMMM (Familial Atypical Multiple Mole-Melanoma)	**Melanoma panel**
☐	Familial pancreatic carcinoma	**PACA panel**
☐	Short stature, basic gene panel	**Growth panel**
☐	Hereditary Multiple Osteochondromas	**HMO panel**
☐	LYNCH syndrome	**LYNCH panel**
☐	Lipodystrophy	**LIPO panel**
☐	Migraine, familial hemiplegic	**FHM panel**
☐	MODY (Maturity Onset Diabetes of the Young)	**Diabetes panel/ MODYScan**
☐	Muscular dystrophies / myopathies	**Muscle panel/ MuscleScan**
☐	Paragangliomas and/or pheochromocytomas	**PGL panel**
☐	Polyglutamin repeat disorders	**PolyQ**
☐	Polyposis coli, adenomatous*	**Polyp panel**
☐	Polycystic kidney disease	**PKD panel**
☐	Skeletal Muscle Channelopathies	**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

Test

☐	Mental retardation or developmental delay, with or without multiple congenitaldefects	☐ CNV analysis (genome wide)
☐	Microdeletion syndrome (specify)	☐ CNV analysis (genome wide)
☐	Growth disorders	☐ CNV analysis (genome wide)
☐	Carrier detection as a result of array finding	☐ CNV analysis (genome wide)

REQUISITION FORM FOR MOLECULAR GENETIC TESTING

Disorder/Referral	Type	Gene/Test
Blood diseases		
☐ Hemochromatosis	Type 1	☐ HFE
☐ Hemoglobinopathies / Thalassemia		
Please use "Requisition form Hemoglobinopathy analysis"		
☐ Hemophilia *(Please send in 2 tubes EDTA blood)*	Type A	☐ F8
	Type B	☐ F9
Cancer genetics		
*Requests only by a consultant clinical geneticist		
☐ Breast- and ovarian cancer, hereditary *		☐ ATM
		☐ BARD1
		☐ BRCA1
		☐ BRCA2
		☐ BRIP1
		☐ CHEK2
		☐ PALB2
		☐ RAD51C
		☐ RAD51D
☐ Clear cell meningioma/ Familial Multiple Meningioma*	CCM	☐ SMARCE1
		☐ SMARCB1
☐ FAMMM (Familial Atypical Multiple Mole-Melanoma)*		☐ CDKN2A
		☐ CDK4
		☐ POT1
		☐ BAP1
		☐ MITF
☐ Gastrointestinal Stromal Tumors (GIST, Carney-Stratakis syndrome)		☐ SDHA
☐ Hyperparathyroidism-jaw tumor syndrome (HPT-JT/HRPT2)		☐ CDC73
☐ Lynch syndrome (HNPCC)*		☐ MLH1
		☐ MSH2 (incl. EPCAM)
		☐ MSH6
		☐ PMS2
☐ Myeloproliferative diseases (MPDs, somatic mutation)		☐ JAK2 (p.Val617Phe)
		☐ MPN-combi: JAK2 exon 12 & exon 14 p.(Val617Phe), MPL exon 10 and CALR exon 9

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

Channelopathies

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

Diabetes

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

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

Growth and skeletal defects

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

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Disorder/Referral	Type	Gene/Test
Immune system		
☐ Chilblain lupus	Type 1	☐ TREX1
☐ Granulomatous disease, chronic, X-linked		☐ CYBB
☐ Lymphoproliferative syndrome, X-linked		☐ XLP
☐ Mediterranean fever, familial (FMF)		☐ MEFV
☐ Wiskott-Aldrich syndrome		☐ WAS
Metabolic diseases		
☐ Adrenal hypoplasia, congenital		☐ NR0B1 (DAX1)
☐ Cystinuria		☐ SLC3A1
		☐ 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) associated with AChR deficiency	Type 9	☐ MUSK
☐ Congenital myasthenic syndrome-10 (CMS10)	Type 10	☐ DOK7
☐ Congenital myasthenic syndrome-11 (CMS11) associated with acetylcholine receptor deficiency	Type 11	☐ RAPSN
☐ 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) *(Please send in 2 tubes of EDTA blood)*	Type 1	☐ Rearrangement chromosome 4
	Type 2	☐ SMCHD1 ☐ LRIF1 ☐ DNMT3B

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☐ Limb Girdle	Myofibrillar myopathy	☐ MYOT
	Emery–Dreifuss muscular dystrophy (EDMD)	☐ LMNA
	Rippling muscle disease	☐ CAV3
	LGMD D4 / R1	☐ CAPN3
	LGMD R2	☐ DYSF
	LGMD R5	☐ SGCG
	LGMD R3	☐ SGCA
	LGMD R4	☐ SGCB
	LGMD R6	☐ SGCD
	LGMD R7	☐ TCAP
	LGMD R8	☐ TRIM32
	LGMD R9	☐ FKRP
	LGMD R12	☐ ANO5
☐ Miyoshi (MMD3)		☐ ANO5
☐ Myopathy with extrapyramidal signs		☐ MICU1

Neurogenetics

☐ Aicardi-Goutières syndrome	Type 1	☐ TREX1
☐ Alternating Hemiplegia of Childhood	Type 2	☐ ATP1A3
☐ CADASIL		☐ NOTCH3
☐ CARASIL/ CADASIL	Type 2	☐ HTRA1
☐ Cerebral hemorrhage with amyloidosis (HCHWA-D)		☐ APP
☐ Dentatorubral-pallidoluysian atrophy (DRPLA)		☐ ATN1
☐ Episodic ataxia	Type 2	☐ CACNA1A
☐ Huntington disease		☐ HTT
☐ Huntington, disease-like 2 (HDL2)		☐ JPH3
☐ Hyperekplexia (familial Startle disease)		☐ GLRA1
		☐ GLRB
		☐ SLC6A5
☐ Migraine, familial hemiplegic (FHM)		☐ ATP1A2
		☐ CACNA1A
		☐ SCN1A
☐ Myoclonus dystonia syndrome		☐ SGCE

Disorder/Referral	Type	Gene/Test
☐ Neuronal ceroid lipofuscinosis (NCL)	Juvenile	☐ CLN3
	Late infantile	☐ TPP1 (CLN2)
	Late infantile	☐ CLN6
	Late infantile	☐ CLN8
	Late infantile / adult	☐ PPT1 (CLN1)
☐ Paroxysmal torticollis		☐ CACNA1A
☐ Polyglutamin repeat disorders		☐ CACNA1A, TBP, ATXN1, ATXN7, ATXN2, ATXN3 and ATN1
☐ Retinal vasculopathy with cerebral leukodystrophy (RVCL)		☐ TREX1

Polycystic kidney disease

☐ Autosomal dominant Polycystic kidney disease (ADPKD)	Dominant	☐ PKD1
	Dominant	☐ PKD2
☐ Autosomal dominant Polycystic kidney and liver disease (ADPKD)	Dominant	☐ GANAB
☐ Autosomal recessive Polycystic kidney (ARPKD)	Recessive	☐ PKHD1
☐ Renal cysts and diabetes syndrome (RCAD)	Dominant	☐ HNF1B

Syndromes

☐ Coffin-Siris syndrome	☐ ARID1A
	☐ ARID1B
	☐ SMARCA4
	☐ SMARCB1
	☐ SMARCE1
☐ Ellis van Creveld syndrome	☐ EVC
	☐ EVC2
☐ Filippi syndrome	☐ CKAP2L
☐ Marshall-Smith syndrome	☐ NFIX
☐ Nicolaides-Baraitser syndrome	☐ SMARCA2

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Disorder/Referral	Type	Gene/Test
☐ Peters Plus syndrome	☐	B3GLCT (B3GALTL)
☐ Pitt-Hopkins syndrome	☐	TCF4
☐ Rubinstein - Taybi syndrome	☐	CREBBP
	☐	EP300
☐ Sotos syndrome	☐	NSD1
☐ Sotos-like syndrome	☐	DNMT3A
	☐	NFIX
	☐	SETD2
	☐	HIST1H1E
☐ TAR (thrombocytopenia-absent radius) syndrome	☐	1q21.1 deletion and RBM8A SNP
☐ Weaver syndrome	☐	EZH2

Other

☐ Hypocalciuric Hypercalcemia, Familial (FHH)	☐	CASR
	☐	GNA11
	☐	AP2S1
☐ Keratosis follicularis spinulosa decalvans (KFSD)	☐	MBTPS2
☐ TSH deficiency and macroorchidism, X-linked	☐	IGSF1