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

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 adress
LUMC, Building 2
KG, Genome diagnostics S-06-P

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

Reply number 10392, 2300 WB Leiden
The Netherlands

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 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.

PATIENT INFORMATION:
Please give to the patient, this can be found at https://www.lumc.nl/over-het-lumc/afdelingen/klinischegenetica/aanvraagformulieren/
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)
☐ 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
☐ Only 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
☐	Basal cell Carcinoma	**BCC panel**
☐	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 CNV 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

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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 ☐ Permissive haplotype analysis (4qA/B)
	Type 2	☐ SMCHD1 ☐ LRIF1 ☐ DNMT3B

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Disorder/Referral	Type	Gene/Test
☐ 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

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