DEPARTMENT OF CLINICAL GENETICS
LABORATORY FOR DIAGNOSTIC GENOME ANALYSIS - LDGA

REQUISITION FORM FOR MOLECULAR GENETIC TESTING

The LDGA 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.

Surname and Initials
Name spouse
Streetname and number
Postal code and city
Country
Date of birth
Sex

Patient information / Fill out completely

Mailadress:
LDGA
LUMC - building 2, Postal zone S-06-P
Einthovenweg 20, 2333 ZC Leiden
P.O. box 9600, 2300 RC Leiden
The Netherlands

Administration:
Tel. : +31 71 5269800
Fax : +31 71 5268276
Email : ldga@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**: 2 EDTA blood tubes (7-10 ml; neonates: 1 tube ≥ 2.5 ml), DNA (2 tubes), tissue, chorionic villi (20 mg) or amniotic fluid (15 ml).
RNA TESTING: Use the "RNA ANALYSIS form".

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

FORM: Please fully complete the form (**one form per person**). The page with patient information **should be given** to the patient!

For diagnostic turnaround times and our current criteria for diagnostic requests, see www.lumc.nl/klingen.

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

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

GENE(S) / TEST: ... **(see next pages for overview)**

Did you previously send us material from this patient, a family member or spouse?
O NO O YES (patient) O YES (family members, fill in table)

Known mutation: yes: …… LDGA Family number (F-No.): ……

CLINICAL INFORMATION and/or **PEDIGREE** (mark the person to be investigated with an 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

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Gene panels	Alias
See next pages for request of individual genes	
○ 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
○ FAMMM (Familial Atypical Multiple Mole-Melanoma)	Melanoma panel
○ Growth disorders	Growth panel
○ LYNCH syndrome	LYNCH panel
○ MODY (Maturity Onset Diabetes of the Young)	Diabetes panel MODYScan
○ Muscular dystrophies / myopathies	Muscle panel MuscleScan
○ Paragangliomas and/or pheochromocytomas	PGL panel
○ Polyposis coli, adenomatous*	Polyp panel

For an overview of all genes in the gene panels see https://www.lumc.nl/org/klinische-genetica/patientenzorg/aanvragen-laboratoriumdiagnostiek/1768733/?setlanguage=English&setcountry=en

NB. NGS is performed by GenomeScan B.V. (with the exception of Melanoma and PGL panel)

Genome analysis	Test
○ Mental retardation or developmental delay, with or without multiple congenital defects	○ Array diagnostics
○ Microdeletion syndrome (specify)	○ Array diagnostics
○ Growth disorders	○ Array diagnostics
○ Carrier detection as a result of array finding	○ Array diagnostics

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Disorder/Referral	Type	Gene/Test
Blood diseases		
○ Hemochromatosis	Type 1	○ HFE
○ Hemoglobinopathies / Thalassemia Please use "Requisition form Hemoglobinopathy analysis"		
○ Hemophilia	Type A	○ F8
	Type B	○ F9
Cancer genetics		
**Requests only by a consultant clinical geneticist*		
○ Breast- and ovarian cancer, hereditary *		○ BRCA1
		○ BRCA2
		○ PALB2
		○ CHEK2 (c.1100delC)
		○ All three above
		○ BRIP1
		○ RAD51C
		○ RAD51D
○ Clear cell meningioma*	CCM	○ SMARCE1
○ FAMMM (Familial Atypical Multiple Mole-Melanoma)*		○ CDKN2A
		○ CDK4
		○ POT1
		○ BAP1
○ Gastrointestinal Stromal Tumors (GIST, Carney-Stratakis syndrome)		○ SDHA
○ Hyperparathyroidism-jaw tumor syndrome, hereditary		○ CDC73
○ Lynch syndrome (HNPCC)*		○ MLH1
		○ MSH2 (incl. EPCAM)
		○ MSH6
		○ PMS2
○ Myeloproliferative diseases (MPDs, somatic mutation)		○ JAK2(p.Val617Phe)
		○ JAK2 (exon 12) and CALR (exon 9)
○ Paragangliomas and/or pheochromocytomas		○ MAX
		○ SDHA
		○ SDHAF2
		○ SDHB
		○ SDHC
		○ SDHD
		○ TMEM127
○ Polyposis coli, adenomatous*	FAP1	○ APC
	MAP	○ MUTYH (incl. GREM1)
	NAP	○ NTHL1
	PPAP	○ POLD1
	PPAP	○ POLE
	FAP4	○ MSH3
○ Renal Cell Carcinoma (RCC), hereditary		○ SDHB
○ Rhabdoid tumor predisposition syndrome (RTPS)*	RTPS1	○ SMARCB1
	RTPS2	○ SMARCA4
○ 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
○ 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 7	○ KLF11
	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
○ 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
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

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Muscular dystrophies		
○ Western blotting (on muscle biopsy)		○ *protein diagnostics*
○ Congenital myasthenic syndrome-11 associated with acetylcholine receptor deficiency (CMS11)		○ RAPSN
○ 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	○ Rearrangement chromosome 4
	Type 2	○ SMCHD1
○ Limb Girdle	Type 1A	○ MYOT
	Type 1B	○ LMNA
	Type 1C	○ CAV3
	Type 2A	○ CAPN3
	Type 2B	○ DYSF
	Type 2C	○ SGCG
	Type 2D	○ SGCA
	Type 2E	○ SGCB
	Type 2F	○ SGCD
	Type 2G	○ TCAP
	Type 2H	○ TRIM32
	Type 2I	○ FKRP
	Type 2L	○ 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
○ 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
○ Neuronal ceroid lipofuscinosis (NCL)	Juvenile	○ CLN3
	Late infantile	○ TPP1 (CLN2)
	Late infantile	○ CLN6
	Late infantile	○ CLN8
	Late infantile / adult	○ PPT1 (CLN1)
○ Paroxysmal torticollis		○ CACNA1A
○ 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
○ 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		
○ Calcemia (hyper/hypo), familial		○ CASR
○ Keratosis follicularis spinulosa decalvans (KFSD)		○ MBTPS2
○ TSH deficiency and macroorchidism, X-linked		○ IGSF1

Leiden University Medical Center
Department of Clinical Genetics

GIVE THIS SECTION TO THE PATIENT

Information for patients regarding the secondary use of tissue

Your biological tissue (e.g. blood, urine, skin, mouth swabs, CVS /amniotic fluid) has been used for chromosomal, DNA or biochemical research for a particular disorder. After completion of diagnostic procedures and testing there is generally a small amount of material remaining that is not simply destroyed. This is referred to as 'residual material'. This residual material is often used for scientific research into your condition, and almost all knowledge about health and disease is acquired through medical scientific research.

This research may occur in several ways, such as through study of a single patient, through the comparison of data from a group of patients with other patients or healthy persons or, frequently, through studies in a research laboratory. In much of this research, residual patient material is used. Use of this material occurs in a coded manner, with the researcher unaware of the identity of the patient and thus unable to directly trace it to a specific individual. Only the person who gave the material to the researcher has a key to the code and is aware of the identity of the treating physician. Within the laboratory one person is designated to apply and carry responsibility for a unique code.

If it is necessary for the research that the researcher knows the identity of those involved - the material is thus traceable - your specific permission is required and this will be requested and discussed with you in advance.

It occasionally happens that a researcher discovers something of direct importance to a particular patient. Should this occur, the person who has the key to the code will inform your doctor, who will then discuss this information with you.

What should you do?

- You do not have to do anything if you do not object to the use of your residual biological material for research in which the researcher does not have access to your personal data.
- If you do object, you can discuss this with your doctor. This will be registered and passed on to the laboratory, so that the residual material is not used.
- If you have no objection and wish to be informed of results important to you or your family, you can also discuss this with your doctor.
- You will be contacted and informed in case of research in which the researcher must have access to your personal details. Your written permission is always needed for this type of research.

We hope that you now have sufficient information. The full text of this brochure is available at www.federa.org. The text and codes of conduct can also be requested from Federa - FMWV (Federation of Medical Scientific Societies). The address is Erasmus MC, JNI WS Ae409, FMWV, PO Box 2040, 3000CA, Rotterdam.