CHKG-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.

TESTEN RvA M007

| | |
|---|---|
| Surname and initials | _____ |
| Name spouse | _____ |
| Street name and number | _____ |
| Postal code and city | _____ |
| Country | _____ |
| Date of birth | _____ |
| Sex | _____ |

**Patient information / fill out completely**


Mail address:
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 71 526 9800
Fax: +31 71 526 8276
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.

| | | | |
|---|---|---|---|
| **REFERRING PHYSICIAN** | : | Telephone | : |
| Hospital/Institution | : | Department | : |
| Address | : | Your ref. no. | : |
| Postal code / City | : | Email | : |

**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 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 LABORATORY:

..-nummer: Datum ontvangst: Paraaf ontvangst:

..-nummer: Hoeveelheid ontvangen bloed:

Familienummer: Paraaf staf:

# 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.

| Gene panels | Alias | Genes |
|---|---|---|
| *See next pages for individual genes* | | |
| o CADASIL and cerebral angiopathies / adult-onset leukoencephalopathies | CHA panel | ABCD1, APP, AUH, CBS, CLCN2, COL4A1, COL4A2, CSF1R, CST3, CTSA, CYP27A1, DARS2, GBE1, GFAP, GLA, GSN, HTRA1, ITM2B, LMNB1, MMACHC, NOTCH3, TREM2, TREX1, TTR, TYMP, TYROBP |
| o Coffin-Siris / Nicolaides-Baraitser syndrome | CSS panel | ARID1A, ARID1B, SMARCA2, SMARCA4, SMARCB1, SMARCE1 |
| o Colorectal carcinoma* | CRC panel | APC, MLH1, MSH2, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, STK11 |
| o FAMMM (Familial Atypical Multiple Mole-Melanoma)* | Melanoma panel | ACD, BAP1, CDK4, CDKN2A, MITF, POT1, TERF2IP, TERT |
| o Growth defects (short stature) | Growth panel | COMP, FGFR3, GH1, GHR, GHSR, IGF1, IGFALS, IGF1R, NPR2, SHOX, STAT5B |
| o MODY (Maturity Onset Diabetes of the Young) | Diabetes panel MODYScan | ABCC8, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1 |
| o Obesity | OBESE panel | ALMS1, ARL6, BBS1, BBS2, BBS4, BBS7, BBS9, BBS10, BBS12, BDNF, CEP290, FTO, G6PC, IRX5, LEP, LEPR, LZTFL1, MAGEL2, MC4R, MKKS, MKS1, NDN, NTRK2, PAX6, PCK1, PCSK1, PHF6, POMC, PPARG, PTEN, SIM1, SNRPN, SPG11, TBX3, THRB, TMEM67, TRIM32, TTC8, WDPCP, WT1 |
| o Paragangliomas and/or pheochromocytomas | PGL panel | MAX, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL |
| o Polyposis coli, adenomatous* | Polyp panel | APC, MUTYH, NTHL1, POLD1, POLE |
| o Muscular dystrophies / myopathies | Muscle panel MuscleScan | ACTA1, ANO5, CAPN3, CAV3, DAG1, DES, DMD, DNAJB6, DYSF, EMD, FHL1, FKRP, FKTN, FLNC, GAA, GMPPB, GNE, ITGA7, LAMA2, LDB3, LMNA, MICU1, MYH7, MYOT, NEB, PLEC, POMGNT1, POMT1, POMT2, SEPN1, SGCA, SGCB, SGCD, SGCG, SMCHD1, TCAP, TPM3, TRIM32, TTN |

**Requests only by a consultant clinical geneticist

| Genome analysis | Test |
|---|---|
| o Mental retardation or developmental delay, with or without multiple congenital defects | o Array diagnostics |
| o Microdeletion syndrome (specify) ..... | o Array diagnostics |
| o Short stature | o Array diagnostics |
| o Carrier detection as a result of array finding | o Array diagnostics |

# 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.

| Disorder/Referral | Type | Gene/Test |
|---|---|---|
| **Blood diseases** | | |
| o Hemochromatosis | Type 1 | o HFE |
| o Hemoglobinopathies / Thalassemia<br>Please use "Requisition form Hemoglobinopathy analysis" | | |
| o Hemophilia | Type A | o F8 |
| | Type B | o F9 |
| **Cancer genetics** | | |
| **Requests only by a consultant clinical geneticist* | | |
| o Breast- and ovarian cancer, hereditary * | | o BRCA1 |
| | | o BRCA2 |
| | | o CHEK2 (c.1100delC) |
| o Clear cell meningioma* | | o SMARCE1 |
| o FAMMM (Familial Atypical Multiple Mole-Melanoma)* | | o CDKN2A |
| | | o CDK4 |
| | | o POT1 |
| o Gastrointestinal Stromal Tumors (GIST, Carney-Stratakis syndrome) | | o SDHA |
| o Hyperparathyroidism-jaw tumor syndrome, hereditary | | o CDC73 (HRPT2) |
| o Lynch syndrome (HNPCC)* | | o MLH1 |
| | | o MSH2 |
| | | o MSH6 |
| | | o PMS2 |
| o Myeloproliferative diseases (MPDs, somatic mutation) | | o JAK2 |
| o Paragangliomas and/or pheochromocytomas | | o MAX |
| | | o SDHA |
| | | o SDHAF2 |
| | | o SDHB |
| | | o SDHC |
| | | o SDHD |
| | | o TMEM127 |
| o Polyposis coli, adenomatous* | FAP | o APC |
| | MAP | o MUTYH |
| | NAP | o NTHL1 |
| | PPAP | o POLD1 |
| | PPAP | o POLE |
| o Renal Cell Carcinoma (RCC), hereditary | | o SDHB |
| o Rhabdoid tumor predisposition syndrome (RTPS)* | Type 1 | o SMARCB1 |
| | Type 2 | o SMARCA4 |
| 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 |
| 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 7 | o KLF11 |
| | Type 9 | o PAX4 |
| | Type 10 | o INS |

| | | |
|---|---|---|
| | Type 11 | o BLK |
| o PNDM (Permanent Neonatal Diabetes Mellitus) | | o 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 / Hereditary Multiple Exostoses | | o EXT1 |
| | | o EXT2 |
| 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 Tall stature | | o NPR2 |
| o Thanatophoric dysplasia | | o FGFR3 |
| o Van Buchem disease | | o VBCH |
| **Immune system** | | |
| o Agammaglobulinemia, X-linked | | o BTK |
| 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** | | |
| o Immunohistochemistry and/or Western blotting (on muscle biopsy) | | o *protein diagnostics* |
| o Duchenne and Becker | | o DMD |
| o Emery-Dreifuss (X-linked) | | o EMD |
| o Facioscapulohumeral (FSHD) | Type 1 | o Rearrangement chromosome 4 |
| | Type 2 | o SMCHD1 |
| o Limb Girdle | Type 1A | o MYOT |
| | Type 1B | o LMNA |
| | Type 1C | o CAV3 |
| | Type 2A | o CAPN3 |
| | Type 2B | o DYSF |
| | Type 2C | o SGCG |
| | Type 2D | o SGCA |
| | Type 2E | o SGCB |
| | Type 2F | o SGCD |
| | Type 2G | o TCAP |
| | Type 2H | o TRIM32 |
| | Type 2I | o FKRP |
| | Type 2L | o ANO5 |
| o Miyoshi (MMD3) | | o ANO5 |
| o Myopathy with extrapyramidal signs | | o MICU1 |

# REQUISITION FORM FOR MOLECULAR GENETIC TESTING



| | | |
|---|---|---|
| **Neurogenetics** | | |
| o Aicardi-Goutières syndrome | Type 1 | o TREX1 |
| o Alternating Hemiplegia of Childhood | | o ATP1A3 |
| o CADASIL | | o NOTCH3 |
| 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 Migraine, familial hemiplegic (FHM) | | o ATP1A2 |
| | | o CACNA1A |
| | | o SCN1A |
| o Myoclonus dystonia syndrome | | o SGCE |
| o Neuronal ceroid lipofuscinosis (NCL) | Juvenile | 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 |
| o Retinal vasculopathy with cerebral leukodystrophy (RVCL) | | o TREX1 |
| **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 |
| o Peters Plus syndrome | | o B3GLCT (B3GALTL) |
| o Rubinstein - Taybi syndrome | | o CREBBP |
| | | o EP300 |
| o Sotos syndrome | | o NSD1 |
| o Sotos-like syndrome | | o DNMT3A |
| | | o NFIX |
| | | o SETD2 |
| o TAR (thrombocytopenia-absent radius) syndrome | | o 1q21.1 deletion and RBM8A SNP |
| o Weaver syndrome | | o EZH2 |
| **Other** | | |
| o Calcemia (hyper/hypo), familial | | o CASR |
| o Keratosis follicularis spinulosa decalvans (KFSD) | | o MBTPS2 |
| o Obesity, juvenile hereditary form | | o LEP |
| | | o LEPR |
| | | o MC4R |
| | | o PPARG |
| o Polycystic kidney disease | Dominant | o PKD1 |
| | Dominant | o PKD2 |
| | Recessive | o PKHD1 |
| o TSH deficiency and macroorchidism, X-linked | | o IGSF1 |

Leiden University Medical Center
Center for Human and Clinical Genetics, **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.