CHKG-DEPARTMENT OF CLINICAL GENETICS
LABORATORY FOR DIAGNOSTIC GENOME ANALYSIS - LDGA

# REQUISITION FORM FOR MOLECULAR DIAGNOSTIC ANALYSIS

The LDGA is NEN-EN-ISO 15189:2007 accredited by the Dutch Accreditation Council. The scope for accreditation number M007 can be found at www.rva.nl.

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 – gebouw 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

**MATERIAL TO SEND:** 2 tubes (7-10 ml, neonates 2 x 2,5 ml) EDTA blood, DNA, tissue, chorionic villi (20 mg) or amnion fluid (15 ml) clearly labeled with name/patient number and date of birth of the patient. Send in prenatal samples and (muscle) tissue **after** consulting the lab.

**TRANSPORT :** Correctly packed in accordance with international regulations. Please note that samples from outside the Netherlands should be sent per express carrier (if transported by air IATA rules apply), to allow for short delivery times.

**REQUISITION FORM:** Fill out **one form per patient** and give the last page (patient information) to the patient.

*For all diagnostics offered our criteria for laboratory requests apply. Visit our website for these criteria and additional information at Patient and care> information for the clinician.*

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

**REASON FOR REFERRAL**

- ○ confirmation/exclusion of clinical diagnosis
- ○ presymptomatic testing
- ○ carrier detection (for recessive diseases only)
- ○ prenatal testing (**only after consultation**)
- ○ testing for family members
- ○ fetal sex determination (**only after consultation**) gestation period: disorder:
- ○ archiving for future testing, reason:

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

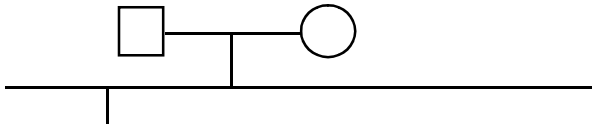
Did you previously send us material from the patient, a family member or spouse?

- ○ **NO**
- ○ **YES** (patient)
- ○ **YES** (family members, fill in table)

Known mutation: yes: ……..

Family number (F-nr):……...

**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:
..-nummer:
Familienummer:
Datum ontvangst:
Hoeveelheid ontvangen bloed:
Paraaf staf:
Paraaf ontvangst:

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Guidelines for turnaround times (TAT):

| | |
|---|---|
| Prenatal testing | 2 weeks |
| Absence/ presence known mutation | 4 weeks |
| Scanning for unknown mutation | 1 – 4 months (see also website) |
| #: Haplotyping | TAT determined after consultation |

| Disorder / Referral | | Gene/Test |
|---|---|---|
| **Blood diseases** | | |
| O Hemochromatosis | | O HFE |
| O Hemoglobinopathies / Thalassemia N.B. Use requisition form Hemoglobinopathy analysis | | |
| O Hemophilia A | | O Factor 8 |
| O Hemophilia B | | O Factor 9 |
| **Diabetes** | | |
| O Hyperproinsulinemia | | O INS |
| O Insulin dependent | | O INS |
| O MIDD (m.3243 tRNALEU/UUR) | | O m.3243 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 10 | O INS |
| O Neonatal, permanent | | O GCK |
| | | O INS |
| | | O KCNJ11 |
| O Persistent hyperinsulinemic hypoglycemia of infancy (PHHI) | | O GCK |
| | | O KCNJ11 |
| **Cancer genetics** | | |
| Δ: All referrals only by a clinical geneticist | | |
| O Breast- and ovariancancer, hereditary Δ | | O BRCA1 |
| | | O BRCA2 |
| O Colorectal cancer, familial adenomatous polyposis Δ | | O APC |
| O Colorectal cancer, hereditary nonpolyposis Δ | | O MLH1 |
| | | O MSH2 |
| | | O MSH6 |
| | | O PMS2 |
| O Colorectal cancer, MUTYH-associated polyposis Δ | | O MUTYH |
| O Melanoma, multiple mole, familial atypical (FAMMM) Δ | | O CDKN2A |
| | | O CDK4 |
| O Pheochromocytomas and/or Paragangliomas | | O MAX |
| | | O SDHAF2 |
| | | O SDHB |
| | | O SDHC |
| | | O SDHD |
| | | O TMEM127 |
| O Hyperparathyroidism-jaw tumor syndrome, hereditary | | O CDC73 (HRPT2) |
| **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 |
| **Genome scan** | | |
| 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 | | O array diagnostics |

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| Disorder / Referral | | Gene/Test |
|---|---|---|
| **Growth and skeletal defects** | | |
| O Achondroplasia | | O FGFR3 |
| O Hypochondroplasia | | O FGFR3 |
| O Langer mesomelic dysplasia (Leri-Weill dyschondrosteosis) | | O SHOX |
| O Osteochondromatosis, multiple (HME) | | O EXT1 |
| O Osteochondromatosis, multiple (HME) | | O EXT2 |
| O Pseudoachondroplastic Dysplasia | | O COMP |
| O Short stature (proportionate) | | O GH1 |
| | | O GHR |
| | | O IGF1 |
| | | O IGF1R |
| | | O IGFALS |
| | | O STAT5B |
| O Thanatophoric dysplasia | | O FGFR3 |
| O Van Buchem disease | | O VBCH |
| **Immune system** | | |
| O Agammaglobulinemia, X-linked | | O BTK |
| O Chilblain lupus | | 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 Systemic lupus erythematosus (SLE) | | O TREX1 |
| O Wiskott-Aldrich syndrome | | O WAS |
| **Metabolic diseases** | | |
| O Adrenal hypoplasia, congenital | | O DAX1 |
| O Cystinuria | | O SLC3A1 |
| | | O SLC7A9 |
| O Hunter syndrome (mucopolysaccharidosis type II) | | O IDS |
| O Ornithine transcarbamylase (OTC) deficiency | | O OTC |
| **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) | | O Rearrangement chromosome 4 |
| O Limb Girdle | Type 1A | O MYOT |
| | 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 With merosin deficiency, congenital | | O LAMA2 # |

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| **Neurogenetics** | | |
| O Aicardi-Goutières syndrome | | O TREX1 |
| O CADASIL | | O NOTCH3 |
| O Dentatorubral-pallidoluysian atrophy (DRPLA) | | O ATN1 |
| O Episodic Ataxia type 2 | | O CACNA1A |
| O Cerebral Hemorrhage with amyloidosis (HCHWA-D) | | O APP |
| O Huntington disease | | O HTT |
| O Huntington,disease-like 2 (HDL2) | | O JPH3 |
| O Hyperekplexia, or familial Startle disease | | O GLRA1 |
| | | O GLRB |
| | | O SLC6A5 |
| O Neuronal Ceroid Lipofuscinosis | Juvenile | O CLN3 |
| | Late infantile | O TPP1 (CLN2) |
| | Late infantile | O CLN6 |
| | Late infantile | O CLN8 |
| | Late infantile, adult | O PPT1 (CLN1) |
| O Migraine, familial hemiplegic | | O CACNA1A |
| | | O ATP1A2 |
| O Myoclonus dystonia syndrome | | O SGCE |
| O Retinal vasculopathy with cerebral leucodystrophy (RVCL) | | O TREX1 |
| **Syndromes** | | |
| O Coffin-Siris syndrome | | O ARID1B |
| O Ellis-van Creveld syndrome | | O EVC1 |
| | | O EVC2 |
| O Fragile X syndrome | | O FMR1 |
| O Marshall-Smith syndrome | | O NFIX |
| O Peters Plus syndrome | | O B3GALTL |
| O Rubinstein - Taybi syndrome | | O CREBBP |
| | | O EP300 |
| O Sotos syndrome | | O NSD1 |
| O Sotos-like syndrome | | O NFIX |
| O TAR (thrombocytopenia–absent radius) syndrome | | O 1q21.1 deletion |
| **Other** | | |
| O Azoospermia/oligozoospermia/infertility (Y-chromosome deletions) | | O AZF genes |
| O Calcemia (hyper/hypo), familial | | O CASR |
| O Keratosis follicularis spinulosa decalvans | | O MBTPS2 |
| O Obesity, early onset | | O MC4R |
| O Polycystic kidney disease, autosomal dominant (ADPKD) | | O PKD1 |
| | | O PKD2 |
| O Polycystic kidney disease, autosomal recessive (ARPKD) | | O PKHD1 |
| O Polycythemia Vera (somatic mutations) | | O JAK2 |
| O Uniparental disomy | | O chromosome … |

Leiden University Medical Center
Center for Human and Clinical Genetics, Department of Clinical Genetics

**Information for patients regarding the secondary use of tissue**

**GIVE THIS SECTION TO THE PATIENT**

**PATIENT INFORMATION**

A sample of your body tissue ( for instance blood, urine, skin, cheek mucous membrane, chorionic villus/ amniotic fluid) has been taken from you for chromosomal, DNA or biochemical evaluation for a particular disorder. After the diagnostic study or test has been done, a small amount of the sample is usually left over, which is not simply destroyed without reason. This is called 'extra' or remaining tissue. The remaining tissue is often usable for scientific research for the same disorder.

Almost all knowledge about health and disease has been gained through medical-scientific research. This research can be conducted in various ways: by examining one patient, by comparing the data of patient groups with that of other patients or healthy persons, and often too by laboratory studies. In much of this scientific research, the remaining tissue from patients is used. This occurs in an encoded fashion: the researcher does not know who the sample has been taken from, so it is not directly traceable to one individual. The only person who has the key to the code, and knows the identity of the referring clinician, is the one who gives the previously collected tissue to the researcher. Within our laboratory, one designated person is appointed and is responsible for that unique code.

If a study requires that the researcher knows who is involved, thus making the body tissue traceable, you need to give your *explicit permission,* and this will be requested and discussed with you in advance.

It can sometimes happen that the researcher discovers something that is of direct importance to a particular patient. In that case the person who has the key to the code will inform the referring clinician. Your doctor will discuss this information with you only if you have indicated that you want to receive such new information.

**What should you do?**

- You don't have to do anything if you *have no objection* to the secondary use of your previously collected tissue for scientific research in which *your personal details are not at the disposal of the researcher.*
- If you *do have an objection*, notify your doctor of this. This information will be registered and passed on to the laboratory, so that the extra tissue will not be used.
- If you have no objection and moreover want to be told about results that are important for you or your family members, you can also inform your doctor of this.
- You will be separately contacted and notified if there is a question of research in which the researcher *must have access to your personal details.* For this type of research your *written permission* is always necessary.

We hope we have provided you with sufficient information. For the complete text of this patient information bulletin, please refer to www.federa.org. You can also request the text and rules of conduct from Federa - FMWV (Federatie van Medisch Wetenschappelijke Verenigingen). The address is Erasmus MC, JNI WS Ae 409, FMWV, PO box 2040, 3000CA Rotterdam.