

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 mls, neonates 2 x 2,5 mls) EDTA blood, DNA, tissue, chorionic villi (20 mgs) or amnion fluid (15 mls) clearly labeled with name/patient number and DOB 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**, the last page with patient information should be given to the patient.

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

| | |
|-----------------------------|----------------|
| REFERRING PHYSICIAN: | Telephone : |
| Hospital/Institution : | Department : |
| Address : | Your ref. ID : |
| Postal code / City : | Email : |

REASON FOR REFERRAL

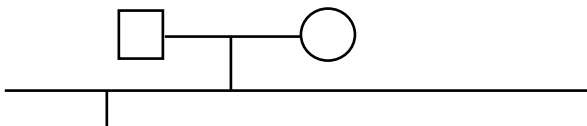
- carrier detection (for recessive diseases only)
- confirmation/exclusion of clinical diagnosis
- presymptomatic testing
- testing for family members
- prenatal testing
- fetal sex determination
- gestation period:
- archiving for future testing, reason:

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):



Previously sent in 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:

Guidelines for turnaround times (TAT):

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

Disease / Referral

Blood diseases

- Hemochromatosis
- Hemoglobinopathies / Thalassemia N.B. Use requisition form Hemoglobinopathy analysis
- Hemophilia A
- Hemophilia B

Diabetes

- MIDD (m.3243 tRNALEU/UUR)
- MODY (Maturity Onset Diabetes of the Young)
 - Type 1
 - Type 2
 - Type 3
 - Type 4
 - Type 5

Cancer genetics

Δ: All referrals only by a clinical geneticist

- Breast- and ovariancancer, hereditary Δ
- Colorectal cancer, familial adenomatous polyposis Δ
- Colorectal cancer, hereditary nonpolyposis Δ
- Colorectal cancer, MUTYH-associated polyposis Δ
- Melanoma, multiple mole, familial atypical (FAMMM) Δ
- Pheochromocytomas and/or Paragangliomas
- Hyperparathyroidism-jaw tumor syndrome, hereditary

Channelopathies

- Hyperkalemic Periodic Paralysis; HYPP
- Hypokalemic Periodic Paralysis; (HOKPP)
 - Type 1
 - Type 2
- Myotonia Congenita; Thomsen, Becker disease
- Myotonia Permanens/fluctuans
- Paramyotonia Congenita

Genome scan

- Mental retardation or developmental delay, with or without multiple congenital defects
- Micro deletion syndrome (specify).....
- Short stature
- Carrier detection

Growth and skeletal defects

- Achondroplasia
- Hypochondroplasia
- Langer mesomelic dysplasia (Leri-Weill dyschondrosteosis)
- Osteochondromatosis, multiple (HME)
- Osteochondromatosis, multiple (HME)
- Pseudoachondroplastic Dysplasia
- Short stature (proportionate)
- Thanatophoric Dysplasia
- Van Buchem disease

Immune system

- Agammaglobulinemia, X-linked
- Granulomatous disease, chronic, X-linked
- Lymphoproliferative syndrome, X-linked
- Mediterranean fever, familial (FMF)
- Wiskott-Aldrich Syndrome

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Disease / Referral

Metabolic diseases

- Adrenal hypoplasia, congenital
- Cystinuria

- Hunter syndrome, Mucopolysaccharidosis type II
- Ornithine Transcarbamylase (OTC) deficiency

Muscular Dystrophies

- Immunohistochemistry and / or Western Blotting
- Muscular dystrophy, congenital with merosin deficiency
- Muscular dystrophy, Duchenne and Becker
- Muscular dystrophy, Emery-Dreifuss (X-linked)
- Muscular dystrophy, Facioscapulohumeral (FSHD)
- Muscular dystrophy, Limb Girdle

- Type 1A
- Type 1C
- Type 2A
- Type 2B
- Type 2C
- Type 2D
- Type 2E
- Type 2F
- Type 2G
- Type 2H
- Type 2I
- Type 2L

Gene/Test

- DAX1
- SLC3A1
- SLC7A9
- IDS
- OTC

- LAMA-2 #
- DMD
- EMD
- Rearrangement chromosome 4
- MYOT
- CAV3
- CAPN3
- DYSF
- SGCG
- SGCA
- SGCB
- SGCD
- TCAP
- TRIM32
- FKRP
- ANO5

Neurogenetics

- CADASIL
- Dentatorubral-pallidoluysian atrophy (DRPLA)
- Episodic Ataxia type 2
- Cerebral Hemorrhage, hereditary, with amyloidosis (HCHWA-D)
- Huntington disease
- Huntington,disease-like 2 (HDL2)
- Hyperekplexia, or familial Startle disease

- Neuronal Ceroid Lipofuscinosis

- Juvenile
- Late infantile
- Late infantile
- Late infantile
- Late infantile, adult

- NOTCH3
- ATN1
- CACNA1A
- APP
- HTT
- JPH3
- GLRA1
- GLRB
- SLC6A5
- CLN3
- TPP1 (CLN2)
- CLN6
- CLN8
- PPT1 (CLN1)
- CACNA1A
- ATP1A2
- SGCE
- TREX1

- Migraine, familial hemiplegic

- Myoclonus- Dystonia Syndrome
- Retinal vasculopathy with cerebral leucodystrophy (RVCL)

Syndromes

- Ellis-van Creveld syndrome

- Fragile X syndrome
- Peters Plus syndrome
- Rubinstein - Taybi syndrome

- Sotos syndrome
- TAR (thrombocytopenia-absent radius) syndrome

- EVC1
- EVC2
- FMR-1
- B3GALTL
- CREBBP
- EP300
- NSD1
- n.a.

Other

- Azoospermia/oligozoospermia/infertility (Y-chromosome deletions)
- Hyper/hypo-calcemia, familial
- Keratosis follicularis spinulosa decalvans
- Obesity, early onset
- Polycystic Kidney Disease, autosomal dominant (ADPKD)
- Polycystic Kidney Disease, autosomal recessive (ARPKD)
- Polycythemia Vera (somatic)
- Uniparental Disomy

- AZF genes
- CASR
- MBTPS2
- MC4R
- PKD1
- PKD2
- PKHD1
- JAK2
- 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.