

REQUEST FORM Lysosomal Storage Disorders (LSD)

Radboudumc

Laboratory for Diagnostics
PO Box 9101
Internal Post 815
6500 HB Nijmegen
The Netherlands

Tel : +31 (0) 24-3614777

www.radboudumc.nl/laboratoriumvoordienst



Investigations are conducted by
the Clinical Genetics Centre
Nijmegen.

Patient

Family name: _____
First name: _____
Middle name: _____
Date of Birth: DD / MM / YY Patient deceased : Yes, date _____
Gender: M / F
Your Reference (MRN etc.etc): _____
Address: _____
ZIP code and City: _____

Referring physician

Name: _____ Phone: _____
Hospital: _____ Fax: _____
Specialty: _____ Email: _____
Department : _____ CC result to: Referring physician
Address: _____ Other: _____
Billing address: _____

Requested Investigations

Please provide clinical data.

Specific test can be ticked on page 2

Previous investigations

Urine accumulation of:
Not yet done / mucopolysaccharides / oligosaccharides: _____

Result deadline

Patient does not give permission for long-term storage for any additional diagnostic or research of this body material at a later date (code 1010)

Specimen

Skin / fibroblasts Date collected _____ Time collected _____

Conditions: Page 2

Other: _____ Date collected _____ Time collected _____

To be filled out by lab employee:

Date received: _____

Reception time: _____

Remarks: _____

Date received:
Time:

Initials
employee:

Specification of clinical data

SPHINGOLIPIDOSES

- Metachromatic leucodystrophy
- GM1-gangliosidosis / Morquio B
- M. Fabry
- Sandhoff / GM2-gangliosidosis
- Tay Sachs / GM2-gangliosidosis
- M. Gaucher
- Niemann-Pick type A/B
- Niemann-Pick type C
- M. Schindler
- M. Krabbe
- Galactosialidosis

MUCOPOLYSACCHARIDOSES

- Hurler/Scheie (MPS I)
- Hunter (MPSII)
- Sanfilippo type A (MPS IIIA)
- Sanfilippo type B (MPS IIIB)
- Sanfilippo type C (MPS IIIC)
- Sanfilippo type D (MPS IIID)
- Morquio A (MPS IVA)
- Maroteaux-Lamy (MPS VI)
- Sly (MPS VII)

MUCOLIPIDOSES

- Mucopolidosis I / Sialidosis
- Mucopolidosis II and III/I-cell disease

OLIGOSACCHARIDOSES

- Fucosidosis
- α mannosidosis
- β -mannosidosis
- Aspartylglucosaminuria

GLYCOGENOSES

- M. Pompe

NEURONAL CEROID LIPOFUSCINOSES

- NCL I
- NCL II

OTHER

- Pseudocholinesterase deficiency
- Lysosomal markerplasma

CATEGORIES

- arylsulfatase A
- β -D-galactosidase A
- α -D-galactosidase
- N-acetyl- β -D-glucosaminidase
- N-acetyl- β -D-glucosaminidase A
- glucocerebrosidase
- Sphingomyelinase
- filipin staining in fibroblasts
- N-acetyl- α -D-galactosaminidase
- galactocerebrosidase
- cathepsin A

- α -L-iduronidase
- iduronate sulfatase
- heparin sulfamidase
- N-acetyl- α -D-glucosaminidase
- acetyl-CoA: glucosaminide N-acetyltransferase
- N-acetylglycosamine-6-sulfate sulfatase
- galactose-6-sulfate sulfatase
- arylsulfatase B
- β -D-glucuronidase

- sialidase (= neuraminidase)
- plasma lysosomal enzymes

- α -L-fucosidase
- α -D-mannosidase
- β -D-mannosidase
- aspartylglucosaminidase

- α -D-glucosidase (= acid maltase)

- palmitoyl-protein thioesterase
- tripeptidyl peptidase

- pseudocholinesterase
- chitotriosidase

Instructions

Shipment of fibroblast cell cultures

When sending a growing fibroblast culture, please completely fill the tube or flask with cell culture medium, and seal the tube/flask by applying Parafilm® around the cap. Please provide us with the passage number, the type of culture medium in which cells were grown, and the antibiotics or fungistats used during cell culture. Cell lines should be free of any infection and should be tested for mycoplasma (e.g. T.R. Chen (1977) in Situ Detection of Mycoplasma Contamination in Cell Cultures by Fluorescent Hoechst 33258 Stain" Exp. Cell Res. 104:255-262).

Please make sure that the sample is protected against damage and against large changes in temperature during transportation. Indicate on the package:

Handle with care; keep at room temperature.

If a courier shipment is used, please inform us by phone (+ 31-24-3614410) or mail (Weefselkweek-TML.LABGK@radboudumc.nl), indicating the waybill number and the name of the courier, for tracking the shipment.

Please make sure that custom forms are filled out properly. It should state: The contents are human cell cultures (or biopsy) for diagnosis (or research, or both), they are non-infectious, have NO commercial value, and will not be used in association with animals. The specimens present no biohazards.

Precautions:

- o Keep at room temperature.
- o Do not expose to X-rays.
- o This parcel may be opened for inspection, but the individual vial should not be opened or the specimen may be disturbed.
- o Courier waybills should state a maximum value for the cells of \$ 10,-- U.S.; also indicate that any customs, duties and taxes will be prepaid with the shipping.

Plasma:

At room temperature if received within 2-3 days by carrier service. Otherwise send frozen.

The sample should be send to:

Radboudumc
Laboratory of Diagnostics
PO Box 9101
Internal Post 815
6500 HB Nijmegen
The Netherlands

Contact:

E-mail: Dirk.Lefebber@radboudumc.nl; Fax: + 31 24 366 8754; Phone: + 31 24 361 4567