

REQUEST FORM Lysosomal Storage Disorders (LSD)

Radboudumc

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

Tel : +31 (0) 24-3614567

www.radboudumc.nl/TML

Translational Metabolic Laboratory

Patient

Family name: _____

First name: _____

Middle name: _____

Date of Birth: DD / MM / YYYY Patient deceased: Yes, date _____

Gender: M / F

Your reference (MRN): _____

Address: _____

ZIP code and City: _____

Referring physician

Name: _____ Phone: _____

Hospital: _____ Fax: _____

Specialty: _____ Email: _____

Department: _____ CC result to: _____

Address: _____

Billing address: _____

Requested investigations

Attention! Please provide clinical data on page 2

Specific test can be ticked on page 2

Previous investigations

Urine accumulation of:

Not yet done / mucopolysaccharides / oligosaccharides:

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	Date collected	Time collected	Protocol after collection
<input type="checkbox"/> Skin / fibroblasts	___/___/___	__:__ hour	Protocol for sending skin biopsy samples ; see also page 2
<input type="checkbox"/> Other: _____	___/___/___	__:__ hour	

To be filled out by lab employee:

Date received: _____

Reception time: _____

Signature employee: _____

Specification of clinical data

Specific investigations

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

- Mucopolipidosis I / Sialidosis
- Mucopolipidosis 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-acetylglucosamine-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 (see also our [protocol](#)):

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 (weefselweek-tml.gen@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 courier service. Otherwise send frozen.

The sample should be send to:

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

Contact:

E-mail: Dirk.Lefeber@radboudumc.nl; Phone: +31 24 361 4567