

## REQUEST FORM Lysosomal Storage Disorders (LSD)

### Radboudumc

Laboratory for Diagnostics  
Internal Post 815  
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The Netherlands

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www.radboudumc.nl/laboratoriumvoordiagnostiek

**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

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: \_\_\_\_\_

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
- $\alpha$ -mannosidosis
- $\beta$ -mannosidosis
- Aspartylglucosaminuria

### GLYCOGENOSES

- M. Pompe

### NEURONAL CEROID LIPOFUSCINOSES

- NCL I
- NCL II

### OTHER

- Pseudocholinesterase deficiency
- Lysosomal markerplasma

### CATEGORIES

- arylsulfatase A
- $\beta$ -D-galactosidase A
- $\alpha$ -D-galactosidase
- N-acetyl- $\beta$ -D-glucosaminidase
- N-acetyl- $\beta$ -D-glucosaminidase A
- glucocerebrosidase
- Sphingomyelinase
- filipin staining in fibroblasts
- N-acetyl- $\alpha$ -D-galactosaminidase
- galactocerebrosidase
- cathepsin A
  
- $\alpha$ -L-iduronidase
- iduronate sulfatase
- heparin sulfamidase
- N-acetyl- $\alpha$ -D-glucosaminidase
- acetyl-CoA: glucosaminide N-acetyltransferase
- N-acetylglucosamine-6-sulfate sulfatase
- galactose-6-sulfate sulfatase
- arylsulfatase B
- $\beta$ -D-glucuronidase

- sialidase (=neuraminidase)
- plasma lysosomal enzymes

- $\alpha$ -L-fucosidase
- $\alpha$ -D-mannosidase
- $\beta$ -D-mannosidase
- aspartylglucosaminidase

- $\alpha$ -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 ([weefselkweek-tml.labgk@radboudumc.nl](mailto: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 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](mailto:Dirk.Lefeber@radboudumc.nl); Fax: +31 24 366 8754; Phone: +31 24 361 4567