

REQUEST FORM Congenital Disorders of Glycosylation (CDG)

Radboudumc

Laboratory for Diagnostics
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6500 HB Nijmegen
The Netherlands

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www.radboudumc.nl/laboratoriumvoordagnostiek



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 etcetc): _____
 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

CDG screening

- Transferrin isofocusing
 Previous results: Not yet done / Type I / Type II
- Apo CIII isofocusing
- Urine oligosaccharides (e.g. MOGS-CDG, NGLY1-CDG)

[Result deadline](#)

CDG subtype determination

- Enzyme analysis (PMM, PMI, PGM)
- N-glycan mass spectrometry
- Exome sequencing
- Targeted gene sequencing [Gene]: _____

- 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 _____
- Heparin plasma Date collected _____ Time collected _____
- DNA Date collected _____
- Other: _____ Date collected _____ Time collected _____

To be filled out by lab employee:

Date received: _____

Reception time: _____

Remarks: _____

Specification of clinical data

Date received:
Time: _____

Initials
employee: _____

Instructions for sample shipment

The laboratory is accredited according EN ISO-15189:2012 (reg M090)
 Chamber of Commerce - Commercial: 41055765 VAT nr.NL8065.52.261.B.01
 Version June 2019

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 courier service. Otherwise send frozen.

The sample should be send to:

Radboudumc

Laboratory for Diagnostics

PO Box 9101

Internal Post 815

6500 HB Nijmegen

The Netherlands

Contact:

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