Radboudumc

REQUEST FORM
Malodour syndrome diagnostics

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

Tel : +31 (0) 24-3614777

www.radboudumc.nl/laboratoriumvoordiagnostiek

Investigations are conducted by the Clinical Genetics Centre Nijmegen.

Patient

Family name: _____

First name: _____

Middle name: _____

Date of Birth: DD / MM / YY

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: _____

Background

I Fish odour syndrome or trimethylaminuria diagnosis

Deficiency of the Flavin-containing Mono-Oxygenase 3 (=*FMO₃*) is responsible for fish odour syndrome or trimethylaminuria (OMIM 602079). First line diagnostics is the mutation analysis of the *FMO₃* gene (requires uncentrifuged EDTA blood or isolated DNA). In case of variants of unknown significance in the *FMO₃* gene we will advise second line testing using a fish meal loading test (a fish meal with preferably 300 grams of fresh fish). In the pre- and post load urine samples we measure trimethylamine (TMA) and the ratio TMA/TMAO using NMR spectroscopy.
NMR spectroscopy also will confirm or exclude dimethylglycine dehydrogenase deficiency as a cause of a fish like malodour.
We prefer to obtain two samples:

1. Urine (>2ml) on normal diet (=pre-load sample)
2. Urine collected during 12 hours after a fish meal with preferably 300 grams of fresh fish. From this volume >2ml should be sent frozen on dry ice (=post-load sample).

II Other malodour syndromes

For other non-fish like malodour syndromes we advise NMR spectroscopy of urine and heparinised plasma (2 ml of each to be sent frozen on dry ice).

Requested Investigations

Please provide clinical data (see page 2).

- **Fish Odour syndrome / Trimethylaminuria**

☐ ***FMO₃*** **gene (first line test)**

☐ **Loading test-fishmeal (second line test)**

(see NMR analysis)

- **Other malodour syndromes**

☐ **NMR spectroscopy in urine, plasma or CSF (first line test)**

(analysis outside scope of ISO15189:2012 accreditation)

☐ 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)

To be filled out by lab employee:

Date received: _____
Reception time: _____
Remarks:

Specimen

- ☐ **EDTA blood (uncentrifuged)** Sample date _____
- ☐ **Urine pre-load** Sample date _____
- ☐ **Urine post-load** Sample date _____
- ☐ **Isolated DNA** Sample date _____

Medication

Please describe the malodour of the patient

Clinical signs and symptoms

Instructions for sample shipment

For DNA analysis:
EDTA 5 ml – do not centrifuge
EDTA plasma for DNA analysis (or isolated DNA) can be sent at room temperature to Radboudumc, Laboratory for Diagnostics, PO Box 9101, Internal Post 815, 6500 HB, Nijmegen, The Netherlands and to the attention of the Translational Metabolic Laboratory.
Urine and heparinized plasma for NMR analysis should be sent well-capped and frozen on sufficient dry ice to Radboudumc, Laboratory for Diagnostics, PO Box 9101, Internal Post 815, 6500 HB, Nijmegen, The Netherlands and to the attention of the Translational Metabolic Laboratory.

For NMR spectroscopy minimal volumes required are:
- Urine; 1 ml
- Heparinized plasma or serum; 1 ml
- Cerebrospinal fluid; 1 ml