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Informed consent for genetic testing in Porphyria

Surname: _____ First name: _____

Date of birth: _____

I confirm that in the context of a genetic counselling session I've been informed about the different aspects of genetic testing.

We suggest molecular-genetic testing in connection with porphyria for you. This disease is an inherited metabolic disorder. We perform molecular-genetic testing that directly detects genetic variants. These variants usually differ for each affected family. With our method we have a greater than 90% chance of detecting them.

Molecular-genetic testing offers you and your family the advantage of precisely detecting variants in your genetic make-up. These results are necessary to

1. evaluate whether or not you or members of your family have inherited porphyria
2. introduce preventive and therapeutic measures
3. evaluate which hereditary risks may result for your descendants

Our testing is limited to the genetic makeup related to porphyria; we will not look at any other aspects of your sample.

You have the right to view the results of the testing at any time and we will be available to discuss the results with you.

Your results and your personal data will be known only to that staff of the Institute for laboratory medicine, which is involved with the testing. No information will be made available to anyone else or any other institution without your consent.

I have understood the information and had sufficient time for decision making.

I give my consent for the following genetic analysis/es:

Incidental findings: Should the analysis/es reveal results not directly related to the testing requested (so called "incidental findings"), I wish to be informed as follow:

- Carrier of a disorder for which preventive and/or therapeutic measures are available ☐YES ☐NO
- Carrier of a disorder for which no preventive / therapeutic measures are yet available ☐YES ☐NO
- Healthy carrier of a recessive disorder which could concern the following generation or other family members ☐YES ☐NO

Other decisions

_____

Should these questions remain unanswered it will be assumed that the patient does NOT want to be informed about incidental findings.

Storage and use of the remaining biological material and data for further analyses.

- I agree that the remaining biological material and data will be stored for possible further analyses. My informed consent will be necessary should further analyses be requested. ☐YES ☐NO

In case of a negative answer the remaining sample will be destroyed!

- I agree that my biological sample and data are used anonymously for quality testing ☐YES ☐NO

Date of birth: _____

The use of your sample and data for research purposes.

Should you agree in principle to participate in research studies you could indicate this below. In a possible scientific publication your data will be made anonymous, in order that no conclusion concerning you can be drawn.Should this be the case you would be contacted at a later stage with details concerning the research projects. A positive answer below is **not yet consent** for the participation in any actual research projects**.**

- In principle, I agree that my biological sample and data could be used for research purposes ☐YES ☐NO

Place and date: _____

Signature: _____

(Patient or parent/legal guardian)

Please inform the following doctor about the results:

Dr. _____

Address: _____

_____

Medical counsellor:

I declare that I've informed the above mentioned person/s, according to the law on genetic testing on humans (GUMG), about the planned genetic tests and their limits as well as providing answers to the patient's questions.

Surname: _____ First name: _____

Signature: _____

Place and date: _____ Stamp: _____

Additional information for the lab:

For familial testing:

First name / Surname / Birth date Indexpatient _____

or Triemli-internal Code Porphyria family _____ Date / Signature _____

New family:

Type of porphyria _____

or Gene _____ Date / Signature _____

or Gene _____ Date / Signature _____