



Diagnostics in suspected cases of Fabry's Disease

Internal laboratory
sample number

Name of patient: _____
 First name: _____
 Date of birth: _____
 Gender: male female
 Address: _____
 Date of sample: _____
 Type of sample: EDTA blood (10ml)

Patient sticker

Sender: Mr. Ms.
Name of physician: _____
 Hospital: _____
 Street: _____
 City: _____
 Tel./Fax: _____

Stamp of facility

Report to: Patient Hospital

Clinical symptoms

- Cardiological symptoms (e.g. cardiomyopathy, arrhythmia)
- Neurological symptoms (e.g. stroke, nonspecific pain)
- Renal symptoms (e.g. proteinuria, renal failure)
- Ophthalmological symptoms (e.g. Cornea verticillata)
- Other suspicious symptoms
 - skin abnormalities (e.g. Angiokeratoma)
 - hypertension
 - cold/heat intolerance
 - nausea / diarrhoe
 - vertigo/hypakusis
 - others

Please use an additional piece of paper to describe the symptoms or family tree if the space provided above is insufficient.

Information:

Please send in at least 10ml (ideally 2x7,5ml) of **unfrozen** EDTA blood for the genetic and biochemical analysis. The samples can be stored in the refrigerator for max 2-3 days. Please remember that the biochemical analysis does not produce any confident results for women; the genetic analysis must always be carried out first for women's samples.

Contact:

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Information regarding genetic Fabry's Disease analysis in accordance with § 9 Genetic Diagnostics Act [Gendiagnostikgesetz (GenDG)]

Name of Patient: **Date of birth:** **Date:**

Your physician has recommended that you undergo a blood analysis for genetic diagnostics in order to determine the presence of a congenital metabolic disorder, Fabry's Disease:

We would like to inform you about the objectives of this analysis, about what occurs during a genetic analysis and what significance the results of such an analysis can have for both you and the members of your family.

The objective of the genetic analysis is to examine your genetic material for any properties which could be the cause of the diagnosed disorder. For this purposes, the analysis of your genetic material (DNA) will be carried out by means of molecular genetic methods. A blood sample (5 – 10 ml) will serve as the **material for analysis**. Usually, a **blood sample** is not associated with any risk to your health. In extremely rare cases it can lead to an accumulation of blood (haematoma) at the site of the puncture or to a damaged nerve.

Significance of the results

The following findings can be the possible outcome of the genetic diagnostics in Fabry's Disease:

- 1) A mutation which causes the disease is discovered. In most cases, this result has a high level of certainty.
- 2) A mutation which has no functional significance is discovered, or a combination of results which does not allow for an accurate diagnosis, creating a need for further examinations. In both cases, a notification with a detailed report and a recommendation regarding the further course will be sent to your attending physician.
- 3) The results do not produce any pathological findings. However, in very rare cases it may be possible that there is an undiscovered mutation in DNA segments which have not been analysed. The presence of Fabry's Disease can thus not be excluded with absolute certainty.

If other members of your family are also being examined, a correct interpretation of the results is dependent on the correct indication of the relationships within the family. In the event that the genetic analysis creates doubt in the indicated relationships within the family, we will only inform you about this if it is crucial to meeting the requirements of our investigation.

The results of the genetic analysis are required and will be used by your attending physicians within the scope of your medical care. A positive test result, i.e. evidence of the presence of Fabry's Disease, is not associated with any risks for you. On the contrary: this allows your physician to offer you and if necessary your family members competent medical advice and better medical care. In contrast to most other genetic disorders there is a licensed method of treatment for Fabry's Disease which contributes to a lasting improvement of many symptoms and can prevent further damage from occurring.

The Genetic Diagnostics Act dictates the **immediate destruction of any samples** following the analysis. Your explicit consent is necessary for a longer storage period of your genetic samples. The same applies if your test results are to be stored for a longer period of time than the statutory retention period of 10 years (e.g. for family testing purposes for hereditary diseases). The transfer of the request for investigation is also dependent on consent. In addition, the examination results may only be passed on to third parties (e.g. other attending physicians, family members) if you give your consent. Of course your personal data and the results of any examinations are subject to **medical confidentiality** requirements, of which only you can release us from.

Storage of the sample material for the purpose of
a possibly necessary or requested checking of the test results
or for additional genetic analyses for diagnostic purposes I agree
 I do not agree

Storage of the sample material for quality control measures regarding
laboratory examinations or for scientific purposes
(please cross out irrelevant option) I agree
 I do not agree

Storage of the results of the analyses for longer than the
statutory retention period of 10 years I agree
 I do not agree

Transfer of the results of the analyses to your attending
physicians (e.g. counselling centres for human genetics, medical
cooperation facilities) I agree
 I do not agree

Right of withdrawal

You can revoke your consent to the analysis either partially or in full at any time without having to indicate a reason. You have the right to not be informed about the results of any examinations (right not to know), to stop any examinations which may have already begun at any time, and to demand that any sample materials as well as any test results up to the time of such a withdrawal be destroyed.

With my signature I also confirm that I was given sufficient time to think about my decision to give consent to the above-mentioned examination.

Date and signature of patient

Stamp and signature of physician

Please keep this copy for your records

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 I do not agree

Storage of the sample material for quality control measures regarding laboratory examinations or for scientific purposes (please cross out irrelevant option) I agree
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Date and signature of patient

Stamp and signature of physician