Patient Information leaflet for clarification / genetic counselling before / after genetic testing according to Gendiagnostikgesetz [Genetic Diagnosis Act]

Dear patient,

You yourself or members of your family possibly suffer from a genetic disorder or you are considering pre-natal diagnosis. Many changes in your genetic make-up can be detected today via the analysis of a suitable sample. Before undergoing genetic testing of any kind, your physician is obliged to inform you about the nature, meaning and consequences of the respective test (duty to inform patient). In case of examination of healthy persons at risk (predictive diagnostics, e.g. in case of neurodegenerative diseases or a hereditary disposition towards cancer) or in case of pre-natal diagnosis, genetic counselling *has* to be offered. You are, however, entitled to refuse genetic counselling in writing if you have already been informed of its contents. The following part will explain to you what exactly genetic counselling comprises and how you can profit from it.

Genetic counselling is designed to help you answer your questions about a possible hereditary disease you (or a family member) might have and to better evaluate the possible consequences (risks) for your life and family plans. In how far genetic counselling can effectively help you, depends on the respective disorder and your personal questions. Both of these factors determine what is discussed within the scope of genetic counselling and how precise statements about genetic risks will be.

Genetic counselling includes <u>regular</u>

- clarification of your personal questions and of the goals the counselling is trying to achieve
- analysis of your personal and familial medical history (anamnesis)
- assessment of available findings and/or reports on findings
- genetic diagnostics conducted as precisely as possible
- extensive information about possible diseases and/or disabilities
- assessment of specific genetic risks
- presentation of the general genetic risks
- extensive counselling concerning the consequences of the abovementioned information for your life and family plans and possibly your health.

Genetic counselling <u>can</u> include

- a physical examination of you or of family members
- taking of samples (e.g. blood, saliva, amniotic fluid, etc.)

Should evidence of additional risks that you had not been aware of turn up in the course of the counselling, these risks will be pointed out. Then, however, it is up to you to decide if and to what extent you would like to be informed and wish to take part in further genetic testing.

Genetic testing will not be conducted without your active agreement to the tests. In these tests, abnormalities might be detected which do not cause damage to your health according to current medical knowledge. You will only be made aware of such abnormalities by us if it is necessary for the conclusion of the examination assignment. An extensive clarification and diagnosis of all possible disorders is not possible, since their causes are often unknown. It is equally impossible to exclude all risk of contracting a disease for you, your family members and especially for your children. In some cases a precise statement about the possibility of contracting a certain disease or disability is impossible. Even if only a small risk (of repetition) is diagnosed, this means that an occurrence is possible. The average probability of congenital diseases and deformities during each pregnancy amounts to approximately 5% (one in twenty).

The most important counselling contents will be documented and if necessary summarized for you in writing. You yourself will decide which doctors are to be informed. Possibly your blood/tissue sample is not analysed at one facility but in several laboratories. In any case, all those involved will observe medical confidentiality and all legal regulations, especially those concerning data protection. Should you have any more questions, please do not hesitate to contact us.

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