Patient information before performing genetic analyses according to the Gendiagnostikgesetz (GenDG) [German Genetic Diagnostics Act]

- to be handed out to the patient -

The German Society of Human Genetics (GfH) and the Professional Society of German Human Geneticists (BVDH) underline that the German Genetic Diagnostics Act requires extensive information to the patient and their written approval for all genetic analyses. Before prenatal and predictive analyses, genetic counselling is required. Please read this informed consent carefully and completely. Please contact us directly if you have any questions.

Dear Patient,

You (or a person in your custody or care) were offered the option to have a genetic analysis performed.

With this information, we want to explain the objective of this analysis and the importance the results may have for you and/or your relatives.

The objective of a genetic analysis is to examine

- the chromosomes as the carriers of the genetic material by means of a chromosome analysis or molecular cytogenetic analysis;
- the genetic material itself (DNA/RNA) by means of molecular genetic analyses or
- the products of the genetic material such as proteins

for genetic characteristics which might be the cause of the disease you or your relatives have, which you or your relative is suspected to have, or for which you or your relative are a carrier and thus being at risk of a certain disease.

In most cases, the test sample is blood (5 ml, frequently less in children). Usually, taking a blood sample is not associated with any risks for your health. In rare cases, an accumulation of blood (haematoma) or damage to a nerve can occur. If a tissue sample (for a skin biopsy, amniocentesis, chorionic villi biopsy or similar procedures) needs to be collected, you will be informed about the respektive risks in a separate patient-doctor discussion.

With a genetic analysis, we are either examining

- specific individual genetic features if a specific suspicion for a disease is indicated; or
- numerous genetic loci at the same time as a screening method (e.g. by means of chromosome analysis, DNA arrays, genome-wide sequencing, RNA sequencing, epigenomic analysis).

Relevance of the results

If a mutation causing a disease is found, these findings are usually highly confirmative of the respective disease. If no mutation causing a disease is found, genetic factors may still be the cause of the disease in the end. Therefore, a genetically determined disease or a predisposition towards a disease may not be excluded with absolute certainty in these unsolved patients. In this case, we will try to determine the probability of the occurrence of the aforementioned disease or a predisposition towards it for you and/ or for your relatives. Sometimes, there is evidence of gene variations for which their clinical relevance is unclear. Due to the enormous number of genetic diseases we cannot inform you about all conceivable (also) genetically determined causes of all diseases. It is also impossible to exclude any risk of all diseases for yourself or your relatives (in particular for your children) by genetic analysis.

In principle, additional results not directly related to the original question can be found with all examination methods (incidental findings). They may be of medical importance for you or your relatives. In particular with screening methods like array analyses or genome-wide sequencing, additional findings can be detected which are indicating an increased risk for a possibly severe disease for which, however, clear management, prevention and/or treatment strategies do exist (so-called actionable genes of which you might not be aware yet). Within the scope of your statement of consent, you can decide whether and/ or under what circumstances you wish to be informed of such incidental findings. However, in some situations, unavoidable additional findings may be observed. If several related persons are examined, accurate interpretation of the findings might depend on the relationship indicated: If the findings or a genetic analysis raises doubt about the indicated relationship, we will tell you only if it is unavoidable in order to fulfil the diagnostic request.

Cancellation policy

You may withdraw your already given consent to this analysis in full or in part at any time without giving any reasons. You are entitled to not receive the results of examinations (right of ignorance), to stop already implemented examination procedures until the time of provision of the report at any time and to demand the destruction of all examination material and findings made up to that time. Already generated reports are not allowed to be destroyed afterwards.

F57/105eng · 03.2023 · UDO

PATIENT IDENTIFICATION STICKER

Institut für Medizinische Genetik und angewandte Genomik
Universitätsklinikum Tübingen

Medical Director
Prof. Dr. med. O. Rieß
Calwerstraße 7
72076 Tübingen

MVZ Fachgebiet Medizinische Genetik
Hoppe-Seyler-Straße 3 · 72076 Tübingen

PATIENT'S STATEMENT OF CONSENT TO A GENETIC ANALYSIS

The **fully completed and signed statement of consent of the patient or their legal representative** is an **essential requirement** for conducting the genetic analysis.

Clinical symptoms / suspected diagnosis / indication / question:

Please delete if **not applicable:**

I hereby consent to the required blood and / or tissue sampling and to the genetic analysis with regard to the above clinical indication.

If necessary, I hereby consent to give the mandate that the diagnostic request can be forwarded to a specialised facility.

I consent to have the diagnostic results and documents being stored for the required period of 10 years.

I consent to have the biomaterial stored for the purpose of verification of the results and additional diagnostic options. I was informed that DNA variations not previously described are documented in a publicly accessible database in anonymised form due to the requirements of the health insurance providers.

I consent that spared biomaterial and the collected data can be used for research purposes and further diagnostic developments in an encoded (pseudonymised) form and that results can be published in scientific journals. For such purpose, I hereby cede the material / the data to the Institute for Medical Genetics and Applied Genomics Tübingen.

If necessary, the results of the examination may be used for counselling and potentially testing members of my family.

Applicable for extensive molecular (cyto-) genetic examinations only:

Within the scope of genetic analyses, information may be obtained that is not directly linked to the diagnostic request but which could be of medical importance for patients and their relatives (incidental findings). Incidental findings may be of immediate clinical importance and be relevant for disease prevention or even for therapy.

[ ] Yes If additional findings will be identified, I wish to be informed about them. **If I do not specifically make a choice about how to handle additional findings, I don´t want to be informed about them.** I do not have a claim to completeness or future updating of such additional findings.

The German Genetic Diagnostics Act restricts the provision of additional findings in prenatal examinations and examinations of persons unable to consent to the examination; therefore, the information may not be passed on despite consent being given. In these cases, the person examined may request the additional findings at a later time (after being deemed capable to consent, usually as of coming of age).

Amendments:

The purpose, type, extent and significance of the diagnostics and in particular the importance and scope of the data were explained to me by the doctor responsible for my care. I was awarded sufficient time for consideration. I may revoke this statement at any time in full or in parts.

Place, date	Signature of the patient/legal representative*	Signature of the doctor providing information	Stamp of the doctor providing information/printed name

(*For children, BOTH parents having custody must give their consent and sign the statement; if not all persons having the care and custody of the child are present, they must have provided a power of attorney.)

Samples to be sent to: Institut für Medizinische Genetik und Angewandte Genomik
MVZ Fachgebiet Med. Genetik
Calwerstr. 7, 72076 Tübingen
Germany

1st page to be forwarded to the Institute for Medical Genetics with the examination request
2nd page to remain with the sender/patient file
3nd page to be forwarded to the patient

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