Health Insurance Company or Cost Unit

Surname, First Name of the Insured Person

Date of Birth

No. Health Insurance Comp. | Insurance No. | Status

Site No. | Physician No. | Date

BIOSCIENTIA HUMAN GENETICS

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Physician Stamp and Signature

Barcode

Molecular Genetic Analyses

Sample Data

- ☐ EDTA blood (3-5 ml, room temperature)
- ☐ DNA (1-5 µg, room temperature)[1]
- ☐ Other

Number of tubes

Sampling date

Time

Biological Gender

- ☐ male
- ☐ female

Invoice Type

- ☐ Statutory health insurance (Referral form sample 10)
- ☐ Private health insurance
- ☐ Self-pay patient
- ☐ Hospital

Clinical Information and Indication

Patient is affected	☐ yes	☐ no	ICD-10-Code:
Family members affected	☐ yes	☐ no	Who:
Parental consanguinity	☐ yes	☐ no	Ethnic origin:

Indication/Suspected Diagnosis/Clinical Details (please provide as much details as possible):

Requested Analysis/Analyses (please tick):

☐ Indication specific analysis of several genes (based on exome/genome sequencing) including copy number analysis (CNVs; if necessary using MLPA; please cross out if CNVs are not desired)
- o Trio analysis*

☐ Single Gene Analysis:
☐ Repeat analysis according to the suspected/differential diagnosis
☐ Methylation analysis according to suspected/differential diagnosis
☐ Segregation of a familial variant (please enclose previous genetic findings):

☐ Optical genome mapping (OGM)** **or** ☐ Chromosomal microarray (CMA)
(for patients with statutory health insurance, CMA/OGM may only be performed after chromosome analysis)
- o Chromosome analysis already performed, result:
- o Please perform chromosome analysis ***

☐ Step-by-step diagnostics for complex cases (e.g. unclear syndromic disorders/developmental disorders); please cross out any analyses not required
1. Chromosome analysis***
2. Analysis of the *FMR1* gene (fragile X syndrome)
3. Optical genome mapping **or** Chromosomal microarray (CMA)
4. Indication-specific trio analysis*

[1] Nucleic acids (DNA/RNA) can only be accepted as primary material if they have been extracted in a CLIA-certified laboratory (comparable to ISO15189) or in a laboratory that fulfills the requirements of CLIA. If you send nucleic acids as primary material, you hereby confirm that the extraction was carried out in an appropriately qualified laboratory.

* Please include blood, consent forms and referral forms from parents
** 2-3 ml EDTA blood, sample must not be older than 4 days, not for prenatal samples
*** Please send 3-5 ml heparin blood and also use the request form "Postnatal chromosome analysis"

BIOSCIENTIA
HUMAN GENETICS

Declaration of Consent for Genetic Testing in Accordance with the Gene Diagnostics Act (GenDG)

I confirm that I have been informed by _____ with regard to the genetic diagnostic test(s) performed on me/the person represented by me _____ described in more detail below according to the German Genetic Diagnostics Act (Gendiagnostikgesetz)

_____

about the purpose, nature, extent, significance and consequences of the requested genetic test(s), the results that can be obtained, the health risks and the intended use of the genetic sample and the test results.

In addition, I confirm that

(1) I have been given sufficient time for consideration before giving consent.

(2) I consent to the test(s) above and the required collection of the genetic sample.

I confirm that I have been informed and I am aware that I can exercise my comprehensive right not to know and that I can also revoke my consent at any time verbally or in writing to the informing physician(s) (responsible medical person), in which case the test will be discontinued and only the service provided up to that point will be billed.

Furthermore, I consent to (Not filling in corresponds to a „no"):

- the storage of the genetic sample after completion of the genetic test(s) so that the laboratory can use it, if necessary, in anonymized form for quality assurance measures and scientific purposes (e.g. statistical evaluations). ☐ yes ☐ no
- the storage of the test results beyond the mandatory period of 10 years, so that they can be used by the laboratory in coded form for quality assurance measures and scientific purposes even after this period. ☐ yes ☐ no
- the communication of medically relevant incidental findings. In case of more comprehensive genetic analyses, depending on the evaluation strategy, variants may be detected by chance which are not related to the indication. However, there is no entitlement to full notification of all incidental findings or future updating of such findings. You have the option to decide whether and which incidental findings are communicated.

  I wish to be informed of incidental findings of:

  - **group 1** (there are preventive or therapeutic measures for a possible illness). ☐ yes ☐ no
  - **group 2** (there are currently no preventive or therapeutic measures for a possible illness). ☐ yes ☐ no
  - **group 3** (variants that can lead to a hereditary disease in offspring or related persons / carriership). ☐ yes ☐ no

  For children and adolescents: Findings of group 1 diseases that manifest in childhood/adolescence will always be communicated. In order to protect the right not to know, group 2 findings are generally not disclosed if the disease only manifests in adulthood and it can be expected that the patient will later be able to give consent.
- the communication of the test results to other attending physicians in the practice/facility or substituting physicians, if my informing physician is not available. ☐ yes ☐ no
- the forwarding of the test request(s) to specialized cooperating laboratories if necessary. In this case, the test results are reported to the laboratory commissioned by me, which is responsible for the further transmission of the results. ☐ yes ☐ no

Place, date Signature of patient or representative

Place, date Signature of physician