Patient data (please fill out clearly in **block letters**)

Family name

First name

Date of birth

Day Month Year

Pat. ID

Age

☐ female

☐ male

0001 160065780060

ABC123S DFÜ-ID: 12345

MOLECULAR GENETIC ANALYSES

BIOSCIENTIA HUMAN GENETICS

Konrad-Adenauer-Str. 17
55218 Ingelheim, Germany
Phone +49-6132-781-240
Fax +49-6132-781-236
E-mail: int.support@bioscientia.com
Website: www.bioscientia.com

Client data

Physician

Sample data

☐ EDTA blood (3-5 ml)
☐ DNA (1-5 µg)
☐ Amniotic fluid
☐ Chorionic villi
☐ Others, please specify

Number of tubes

Sampling date

Time

* Nucleic acid will only be accepted as a specimen type, if extraction is performed in a CLIA-certified laboratory or a laboratory that meets equivalent requirements

Patient is affected ☐ yes ☐ no

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:

☐ Sequencing
- ☐ Indication specific gene panel (based on exome/genome sequencing)
- ☐ Repeat analysis (if indicated/applicable)
- ☐ Single gene analysis:
- ☐ Segregation of a familial variant (please include previous genetic report):

☐ Copy number analysis (CNVs, deletions/duplications)

☐ Array-CGH (180k) **or**

☐ Optical genome mapping (Bionano Genomics, not for prenatal samples)
- ☐ Please also perform chromosome analysis (3-5 ml heparin blood attached**)

☐ Stepwise diagnostic approach for complex cases
(e.g. unclear syndromic disorder/unspecific developmental delay)

1. Chromosome analysis**
2. Analysis of the *FMR1* gene (fragile X syndrome, if indicated)
3. Array CGH **or** optical genome mapping
4. Indication specific trio exome sequencing
(please also send EDTA blood of the parents)

** Please also use request form „Postnatal Chromosome Analysis" and send 3-5 ml heparin blood.

specimen material edta ABC123S2

specimen material edta ABC123S2

specimen material amniotic fluid ABC123SR

specimen material amniotic fluid ABC123SR

specimen material ABC123S5

specimen material ABC123S5

ABC123S

This label should be stuck onto the copy attached and kept for your records.

DO NOT SEND TO US.

ABC123S DFÜ-ID: 12345

These labels are for the patient´s tube(s). Those labels not needed - please tear off and discard - do not send to us.

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)

Exact description of examinations / indications / questions / suspected diagnosis, if applicable

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), 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 notification of any incidental findings. In the course of genetic analyses, information may be obtained that is not directly related to the requested test, but may nevertheless be of medical significance to me or my family (as recommended by the American College of Medical Genetics and Genomics, ACMG, Miller DT et al., 2022: PMID: 35802134; DOI: 10.1016/j.gim.2022.04.006). ☐ yes ☐ no

There is no entitlement to notification of all incidental findings or to future updating of such findings. In the case of minors, there will be no notification of incidental findings for diseases that occur in adulthood. The complete gene list of the ACMG may only be requested by genetic counsellors or physicians with the additional qualification in medical genetics.

- 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 patient or representative

_____

Place, date — Signature physician