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

Family name

First name

Date of birth

Day Month Year

Id. No.

Age

☐ male

☐ female

0001 160065660048

CHROMOSOME ANALYSIS POSTNATAL

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 type

postnatal

☐ Blood / heparin tube

☐ Skin biopsy

☐ Slides / fixed cells

Sampling date:

Time:

No. of tubes sent

Chromosome analysis requested[1]:

☐ Chromosome analysis

☐ Newborn ☐ Suspected mosaicism

☐ Metaphase FISH (microdeletion syndromes)

☐ Cri-du-Chat syndrome

☐ DiGeorge/Velocardiofacial syndrome

☐ Miller-Dieker syndrome

☐ Prader-Willi/Angelman syndrome

EDTA blood required, please use request form "Molecular genetic analyses"

☐ Smith-Magenis syndrome

☐ Williams-Beuren syndrome

☐ Wolf-Hirschhorn syndrome

☐ Others (probes on request)

☐ Chromosomal microarray (CMA)

EDTA blood required, please use request form "Molecular genetic analyses"

☐ Tumor genetics

Please use the "Interdisciplinary blood cancer diagnostics" request form

Clinical data and indication:

☐ Multiple congenital anomalies

☐ Developmental delay

☐ Mental retardation

☐ Dysmorphic features

☐ Habitual miscarriages

☐ Growth retardation

☐ Infertility

☐ IVF/ICSI planned

☐ parental chromosome analysis following abnormal results of a prenatal/postnatal analysis (please specify):

☐ other clinical comments (please specify or attach relevant reports):

[1] These examinations require preparation and analyses of multiple slides. If prepared material contains no mitoses, a handling fee will be charged.

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 of 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, publication in peer-reviewed scientific journals). ☐ 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

Preanalytics and Samples

Heparin blood:	**Infants and small children:** 2-3 ml / room temperature **Adults:** 5-7 ml / room temperature
Cell culture:**	min. 25 ml / room temperature
Fibroblasts / skin biopsy*:**	approx. 5 mg piece of tissue, 2-10 mm^3 in size
EDTA blood (Chromosomal microarray):	5-7 ml / room temperature

** in culture flask, densely grown
*** Use a sterile container with physiological saline solution (0,9% NaCl) that is explicitly suitable for cell transport.