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

Family name

First name

Date of birth

Day Month Year

Id. No.

Age

0001 160065190084

PRENATAL DIAGNOSTICS

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

- ☐ Amniotic fluid
- ☐ Chorionic villi
- ☐ Umbilical cord blood
- ☐ Product of conception
- ☐ EDTA blood

Sampling date **Time** **No. of tubes sent**

Chromosome analysis

- ☐ **Chromosome analysis**
- ☐ **Chromosome analysis and QF-PCR** (Rapid aneuploidy screening)
  Exclusion of aneuploidy of chromosomes 13, 18, 21, X, Y (only in combination with conventional chromosome analysis)
- ☐ **Chromosome analysis and Metaphase FISH**
  - ☐ Exclusion of cryptic translocation
    - ☐ Trisomy 13
    - ☐ Trisomy 18
    - ☐ Trisomy 21
  - ☐ Prader-Willi / Angelman syndrome
  - ☐ Miller-Dieker syndrome
  - ☐ DiGeorge / Velocardiofacial syndrome
  - ☐ Smith-Magenis syndrome
  - ☐ Williams-Beuren syndrome
  - ☐ Cri-du-Chat syndrome
  - ☐ Wolf-Hirschhorn syndrome
  - ☐ Others

Biochemistry

- ☐ **From amniotic fluid**
  - ☐ AFP
  - ☐ AChE

Chromosomal microarray (CMA)

- ☐ **CMA** (indicated in case of an abnormal ultrasound or positive family history for chromosomal changes)
  Please provide EDTA blood from both parents and informed consent for parental CMA if needed.

Molecular genetic analysis

- ☐ **Analysis**

Please specify name of the analysis and give further information (indication, family history, previous reports) or please use our request form "Molecular Genetic Analyses".

Clinical data

Number of fetuses

Last menstrual period (LMP)

Gestational age (Ultrasound) +

Indication

- ☐ Request of the patient
- ☐ Advanced maternal age
- ☐ Abnormal ultrasound finding
- ☐ Abnormal maternal serum screening
- ☐ Previous pregnancy or child with
  - ☐ chromosome anomaly:
    Please enclose previous reports.
  - ☐ Genetic disease:
    Please enclose previous reports.
- ☐ Abnormal results of previous chromosome analyses

Additional information

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

Important information

Prior to a prenatal genetic test, and again after the corresponding results have been determined, the pregnant woman shall receive counselling and shall be advised of her right to counselling (Deutsches Gendiagnostikgesetz (GenDG), Bundesgesetzblatt 2009, Teil I Nr. 50: 2529-2538).

Preanalytics and transport material

Prenatal genetic analyses are performed on:

Chorionic villi: (beginning from the 11th pregnancy week)	- minimum amount of 30 mg (incl. array CGH: 50 mg) - send in a sterile-coated tube with 10 ml physiological NaCl solution - storage and transport at room temperature
Amniotic fluid: (beginning from the 15th pregnancy week)	- 10-15 ml (incl. CMA: 15-20 ml); discard the first 2 ml, **please do not centrifuge!** - storage and transport at room temperature
Umbilical cord blood: (beginning from the 20th pregnancy week)	- 2-3 ml Lithium-Heparin blood as well as 1-2 ml EDTA blood (if molecular genetic analyses/CMA is requested) - storage and transport at room temperature
Product of conception:	- chorionic villi, parts of the umbilical cord, achilles tendon, fascia lata **(material should not be older than 3 days)** - send in a sterile-coated tube with 10 ml physiological NaCl solution - storage and transport at room temperature

To exclude the most common aneuploidies (of chromosomes 13, 18, 21, X and Y), **a prenatal rapid aneuploidy screening (QF-PCR)** can be performed on the amniotic fluid specimen. For this purpose, please remove additional 3 ml amniotic fluid.

Please do not freeze samples. Do not send in formalin!

Please order request forms, shipping supplies and transport materials at Bioscientia International: int.support@bioscientia.com