

Patient data (please fill out clearly in block letters)

Family name

First name

Date of birth

____/____/____

Day Month Year

Id. No.

Age

- male
- female



Request form

PRENATAL DIAGNOSTICS



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
- Slides / fixed cells
- EDTA blood

Sampling date

____/____/____

Time

____:____

No. of tubes sent

Chromosome analysis

- Chromosome analysis
- Chromosome analysis and Interphase FISH (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

Array-CGH

- Array-CGH (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 array-CGH 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
 - Free trisomy 21
 - Translocation trisomy 21
 - Trisomy 21 (without further indications)
 - Trisomy 13
 - Trisomy 18
 - Other chromosome anomaly:

Please enclose previous reports.

- Abnormal results of previous chromosome analyses

Declaration of Informed Consent

- Please delete as appropriate -

With my signature I declare that I was briefed on _____ by the physician:

about the nature, importance and implications of the genetic test and that I give my consent to the genetic analysis marked above and to the collection of blood and tissue samples needed for this purpose. I consent to the results of the tests being made available to the following persons in addition to the doctor who submitted them:

I consent to the storage, in accordance with legal requirements, of the recorded data in paper and/or electronic

form and to their use and/or publication in pseudoanonymized form for scientific purposes or for quality assurance including submission of the results to public databases to advance the understanding of the relationship between genetic changes and clinical symptoms in the medical community. Confidentiality is maintained. I agree that, contrary to legal requirements, my test results and the corresponding report will not be destroyed after 10 years (to enable my family to access these data in the event of my death).

I consent to the communication of my data to a medical billing clearing house for invoicing purposes. I am aware that I may withdraw this consent at any time, verbally or in writing, without giving reasons and without this having any adverse consequences for me.

Following paragraph refers to array-CGH testing:
I am aware that the following results will not be communicated: incidental findings like carriership (for recessive diseases

or X-linked recessive diseases in female fetuses), risk factors and late-onset diseases of the fetus which do not have medical consequences in childhood and adolescence. Furthermore, I am informed that unclear and likely benign copy number changes will not be reported. Additionally, I understand that in case of likely clinically relevant changes in the fetus parental testing needs to be performed for further clarification which requires separate declarations of informed consent of each parent.

Place, date: _____

Name of patient / legal representative _____

Signature of patient / legal representative _____

Please tear off this strip before sending.

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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)	<ul style="list-style-type: none">- minimum amount of 30 mg (incl. array-CGH)- send in sterile transport medium⁺- storage and transport at room temperature
Amniotic fluid: (beginning from the 15th pregnancy week)	<ul style="list-style-type: none">- 10-15 ml (incl. array-CGH: 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)	<ul style="list-style-type: none">- 2-3 ml Lithium-Heparin blood- storage and transport at room temperature
Product of conception:	<ul style="list-style-type: none">- chorionic villi, parts of the umbilical cord, achilles tendon, fascia lata (material should not be older than 3 days)- send in sterile transport medium⁺- storage and transport at room temperature

⁺Transport medium is available at Bioscientia Center for Human Genetics; alternatively, please use a sterile-coated tube with physiological NaCl solution and antibiotics (Penicillin/Streptomycin (100IU/100µg/ml) or Gentamycin (50µg/ml)).

To exclude the most common aneuploidies (of chromosomes 13, 18, 21, X and Y), a **prenatal rapid aneuploidy screening (Interphase FISH)** 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 sent in formalin!

Please order request forms, shipping supplies and transport materials at Bioscientia Center for Human Genetics Ingelheim, Germany.