

ANALYTICAL SPECTRUM

CYTOGENETICS/CHROMOSOME ANALYSES

prenatal:

- amniotic fluid
- chorionic villi (CVS)
- products of conception (POC)
- percutaneous umbilical blood

postnatal:

- blood
- skin biopsy
- bone marrow
- leukemic blood

special testing:

- high resolution banding (prometaphase)
- mosaic chromosome study
- banding (CBG, QFQ, NOR, etc.)
- Fluorescence in situ Hybridization (FISH)

MOLECULAR CYTOGENETIC/ FLUORESCENCE IN SITU HYBRIDIZATION

(List of probes on request)

- Interphase FISH
prenatal rapid aneuploidy screening (only together with a conventional chromosome analysis),
mosaic studies
- chromosome specific/regional specific analysis,
e.g. for all human subtelomer regions
- syndrome specific analysis (microdeletions):
all commercially available probes,
e.g. for Prader-Willi/Angelman, Miller-Dieker,
DiGeorge/Velocardiofacial, Smith-Magenis,
Williams-Beuren, Cri-du-Chat, Wolf-Hirschhorn
syndrome

SOLID TUMOR/ASCITES CHROMOSOME ANALYSES

- bone marrow
- leukemic blood

BIOCHEMISTRY

- Triple Test
- First trimester screening
(certified by the Fetal Medicine Foundation)
- Amniotic fluid alpha-fetoprotein (AFAFP)
- Amniotic fluid acetylcholinesterase (ACHE)

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DNA ANALYSES

- Achondroplasia
- Acute lymphoblastic leukemia
- Alpha-1-antitrypsin deficiency
- Angelman syndrome
- Angiotensin I converting enzyme, Genotyping
- Aniridia
- Apert syndrome
- Apolipoprotein B, Genotyping/Mutation analysis
- Apolipoprotein E, Genotyping/Mutation analysis
- Azoospermia factor
- Beckwith-Wiedemann syndrome
- Breast Cancer
- Coagulation factor II (Prothrombin), Genotyping
- Congenital adrenal hyperplasia (21-hydroxylase deficiency)
- Craniosynostosis
- Chronic myelocytic leukemia
- Crouzon syndrome
- Cystic fibrosis
- Denys-Drash syndrome
- Dystonia, Dopa-responsive
- Ehlers-Danlos syndrome type VII A/B
- Factor V mutation
- Familial Mediterranean Fever
- Fragile X syndrome
- Hemochromatosis
- Hypochondroplasia
- Jackson-Weiss syndrome
- Metaphyseal chondrodysplasia, Schmid type
- Multiple endocrine neoplasia (MEN1 and MEN2)
- Muscular Dystrophy type Becker and Duchenne
- Pfeiffer syndrome
- Prader-Willi syndrome
- Thanatophoric dysplasia
- Torsion Dystonia
- Thyroid hormone resistance
- Vas deferens, congenital bilateral aplasia
- Von Hippel-Lindau syndrome
- Waardenburg syndrome type I/III and type II
- Wilms tumor

more than 300 further analyses on request

SERVICE

TURN – AROUND - TIME

The average turn-around-time of prenatal analysis is around 8 days. The turn-around-time of postnatal analyses and DNA analyses depends on the indication and therefore on the complexity of the analysis (average for chromosome analyses is 9 days; please refer to the special list of all such tests offered by bioscientia).

The results will be reported directly by mail as well as by fax.

QUALITY

The bioscientia Center for Human Genetics is accredited by the „College of American Pathologists (CAP)“ and participates in quality assurance programmes of the German „Association for Medical Genetics“.

We support the request of this organisation to offer sufficient, qualified genetic counselling in conjunction with human genetic testing.

CUSTOMER SERVICE

Sampling and mailing material is available free of charge on request from Bioscientia directly or from one of our local agents.

Before sending samples to the laboratory, please contact us in order to ensure all requirements are met.

Feel free to contact us, if you have any questions.

Do you have further questions concerning our analytical spectrum or services?

We offer a variety of medical leaflets. Please contact us with your request:

HEALTHCARE PROFESSIONAL INFORMATION:

- Interphase FISH – Prenatal rapid aneuploidy screening
- Prenatal diagnosis – Chromosome analyses, technical information
- Diagnosis of mental retardation with unknown cause – Multitelomer/Subtelomer-FISH screening
- Prenatal screening for Down syndrome (Triple test, First trimester screening)
- Breast cancer (BRCA1 and BRCA2)
- Pharmacogenetics – State of the art solutions in modern genetic diagnostics
- Molecular diagnosis of Cystic fibrosis. Special mutation panel for the Middle East
- Analytical spectrum/Human genetic analyses (more than 300 analyses on request)
- Analytical spectrum human genetics: References for sampling and turn-around-times
- Test List General / ___ copies

PATIENT INFORMATION:

- How accurate are the results of prenatal testing
- Prenatal screening for Down syndrome (Triple Test, First trimester screening)
- Genetic counselling

Please contact me:

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NOTES:

bioscientia
CENTER FOR
HUMAN
GENETICS



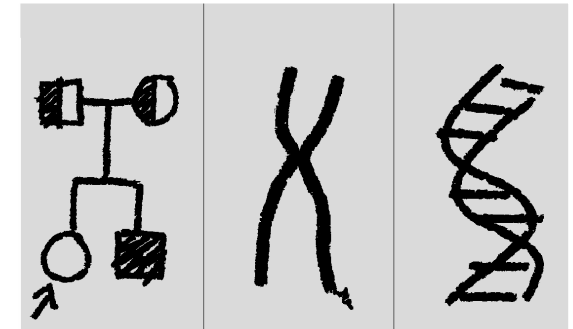
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SERVICES



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