MGZ

Medical Genetics Center

Clinician Information

GENETIC PRENATAL DIAGNOSTICS

Prenatal diagnosis offers a wide range of examination methods that ensure that fetal malformations and genetic diseases of the child can be ruled out or detected early. This also makes genetically complex prenatal disease patterns, which are associated with multiple organ malformations, accessible for meaningful genetic diagnostics.

The entire spectrum of diagnostic tests and methods is available at the MGZ – Medical Genetics Center (MGZ) in order to ensure adequate patient care in the prenatal situation. In addition, timely and adequate logistics and fast sample processing are of great importance. Our medical team is also at your patients' disposal for genetic counseling or for your direct consultation to discuss diagnostic procedures and further patient care.

NON-INVASIVE PRENATAL TESTING (NIPT)

The MGZ offers non-invasive prenatal testing (NIPT) to screen for fetal autosomal (chromosomes 13, 18, and 21) and sex chromosome aneuploidies, by analyzing cell-free fetal DNA in the maternal blood, using Next-Generation-Sequencing.

We take considerable care of pregnant women both pre- and post- test results. Especially in case of positive results, our medical experts are available to discuss further procedures in the context of the individual patient with the patient's responsible doctor.

INVASIVE PRENATAL DIAGNOSTICS

In the case of an abnormal NIPT, or in the case of an abnormal ultrasound, the MGZ offers the following tests/methods for prenatal diagnostics:

Prenatal Chromosome Analysis and Rapid Prenatal Aneuploidy FISH

The rapid prenatal aneuploidy FISH test determines the number of chromosomes 13, 18, 21, X and Y from amniotic fluid within 24 hours. After a chorionic villus sampling, a result on the number and coarse structure of all chromosomes is available one working day after sample receipt. A conventional chromosome analysis examines numerical and structural changes of all chromosomes and is used for confirmatory analysis, e.g. in the case of conspicuous NIPT.

Prenatal Microarray Diagnostics

This is a high-resolution, SNP microarray-based determination of unbalanced changes in the fetal karyotype with a significantly higher detection rate of chromosomal changes compared to conventional chromosome analysis, especially in the case of conspicuous or borderline first trimester tests and ultrasound abnormalities.

Prenatal Targeted Variant Sequencing

Targeted variant sequencing following chorionic villus sampling (CVS) or amniocentesis is possible with a rapid turnaround time, upon prior request. Candidates for this testing are women with a known risk status for a Mendelian disorder. Additional testing for maternal cell contamination (MCC) in the prenatal sample is recommended. Therefore, in parallel with the fetal sample, an EDTA blood sample of the pregnant mother should be provided.

Next-Generation Sequencing (NGS) Analyses

In case of an abnormal fetal ultrasound finding suggestive of a monogenic fetal anomaly syndrome, Next-Generation Sequencing (NGS) is the state-of-the-art molecular diagnostic test to rapidly and efficiently identify a causal genetic variant.

- **Phenotype-based gene panel analyses** can be used to analyse a number of candidate genes in parallel in case of a clinically suggested phenotype (e.g. arthrogryposis, heart defects, kidney, or brain malformations).
- For complex fetal phenotypes, it is also possible to perform a prenatal clinical exome analysis by targeting about 4000 genes associated with known clinical phenotypes.
- Trio Exome Analysis provides improved diagnostic information. In addition to the fetus, the genotypes of non-affected parents are included in the analysis. Trio-based exome sequencing has proven to be an effective diagnostic strategy, especially for the identification of de novo variants in the fetus.

You are welcome to discuss these diagnostic procedures with one of our specialists at **+49 89 3090886-0 (CET)**.

SAMPLE REQUIREMENTS, SHIPPING, AND TURNAROUND TIMES

Sample Requirements

- **NIPT:**
  Special collection requirements. Please contact: **inquiry@mgz-muenchen.com**
- **Chromosome Analysis/Microarray:**
  Amniotic fluid, Chorionic villi (CVS) / Placental villi, Heparinized fetal umbilical cord blood
- **Rapid Prenatal Aneuploidy FISH:**
  Amniotic fluid
- **NGS (fetal sample):**
  Chorionic villi (CVS) / Placental villi, amniotic fluid*, EDTA-stabilized fetal umbilical cord blood – when sending a fetal sample for NGS, we also recommend sending EDTA blood from the parents.
- **Trio-Exome:**
  In addition to the fetal sample, EDTA blood from the parents.

Our team from the cytogenetics department will be happy to answer your questions about the sample requirements at **+49 89 3090886-550 (CET)**.

* Cultivation of the amniotic fluid before DNA extraction is often necessary

Sample Shipping

Prenatal samples can be sent by post or courier to the MGZ. Please contact us at **inquiry@mgz-muenchen.com** if you require assistance with logistics.

- **Shipping Address** (sample receipt Monday - Friday only):

  MGZ – Medical Genetics Center
  Bayerstr. 3-5, Receiving Department: Schlosserstr. 4
  D-80335 Munich
  GERMANY

Turnaround Times

Please contact us in advance of sending prenatal samples by email at **inquiry@mgz-muenchen.com** or by telephone at **+49 89 3090886-653 (CET).** We endeavour to process prenatal samples in our laboratory as quickly as possible.

- **NIPT:** 4 - 6 Working days
- **Rapid Prenatal Aneuploidy FISH:** 1 Working day
- **Chromosome Analysis:** 10 - 14 Working days
- **Microarray, NGS:** Max. 10 Working days
- **Trio Exome:** 10 - 14 Working days

IMPORTANT CONTACT NUMBERS

- **Consultation with our medical team**
  +49 89 3090886-0 (CET)
- **Sample Requirements / Cytogenetics Department**
  +49 89 3090886-550 (CET)

PRICE INQUIRIES AND TEST ORDERING

You can obtain a cost statement for a specific analysis using our convenient website contact form or submit your price inquiry via the MGZ's online portal at **www.mgz-muenchen.com**.

To send a test request, please complete a test request form and patient consent form for each patient and submit these with the samples. All forms are available on our website at **www.mgz-muenchen.com**.

For detailed price and ordering information, including shipping instructions, please see the *How to Order* section of our website.

Still have questions? Contact us at **inquiry@mgz-muenchen.com**.

On our website you will find information on other clinical topics as well as information about our company and services.

Visit us at www.mgz-muenchen.com

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