



Medical Genetics Center

A panoramic view of a city, likely Munich, Germany, featuring a prominent cathedral with two large green domes (St. Mary's Church) and a red-tiled roof. In the background, there are snow-capped mountains under a clear blue sky. The foreground shows residential buildings and greenery.

*Experience how the right clinical partner
can make a difference to your genetic testing needs*



ABOUT US

An established medical practice and laboratory under one roof



Prof. Dr. Elke Holinski-Feder, MD
Clinical Geneticist



PD Dr. Angela Abicht, MD
Clinical Geneticist

Since the year 2000, the MGZ – Medical Genetics Center has been providing **genetic counseling** and **diagnostic genetic testing** services with the highest professional competence in many medical specialties of human genetics. Both local and international clinicians have worked with the MGZ – a reliable and respected provider of genetic services in the diagnosis of **rare diseases and hereditary cancers**.

Our team of medical specialists provides **personalized genetic counseling** to approximately 4000 local and international patients per year and genetic **consultation support** to fellow clinicians on a daily basis. At our on-site **nationally accredited medical genetics laboratory** in Munich, Germany, we perform over **20,000 molecular genetics and cytogenetic analyses, annually**, using state-of-the-art analytical methods.

Our **multi-disciplined medical and scientific team** works in close collaboration in the interpretation of sequence variants, combining in-depth clinical expertise and scientific knowledge, within the clinical context of the individual patient, to provide detailed and informative **clinically actionable reports** to assist clinicians in providing optimal patient care.

Approaching nearly 20 years of experience, with a growing team of now 125 employees, the MGZ is committed to providing clinicians and their patients with **trustworthy, high-quality genetic diagnostics and dedicated clinical and logistical support** throughout the entire diagnostic process.

We look forward to the opportunity of working with you and helping you to experience the difference we can make to your medical genetic testing needs and, ultimately, to making a contribution to the improved care of your patients.

Human Genetics by Clinicians for Clinicians

THE RIGHT CLINICAL PARTNER

Medical responsibility & Expertise you can trust

Assured Competency Through Specialized Training

Our clinical geneticists are medical doctors who have completed a minimum of five years specialized training in the area of clinical genetics and genomics, which covers the full range of clinical diagnosis, knowledge of laboratory techniques, interpretation of data, genetic counseling, and medical management of genetic conditions. In addition, they have different backgrounds in a primary care specialty such as neurology, internal medicine or pediatrics.

A Direct Contact Partner for Your Clinical Needs

For specific clinical and differential diagnostic questions, whether related to a testing strategy or to a particular patient case or test result, our clinical area specialists are your direct contact persons. This peer to peer consultation is available whether genetic testing is imminent or has already been performed through our center or, externally, by another clinical laboratory.

Genetic Counseling/Consultations by our Team of Experts

On a daily basis, our team of experts provides genetic counseling to both patients, and clinicians directly involved in patient care. We assess individuals with genetic disorders, birth defects and cancer, or a possible predisposition to a hereditary disease and counsel them and their families. For these patients, it is our mission to help find the answers – to assist them through their diagnostic journey. In collaboration with fellow clinicians, we discuss possible differential diagnoses in order to design a testing strategy best suited to the patient.



Detailed Interpretation of the Results

The results from the genetic analyses are always interpreted in the context of the patient, the initial clinical question, and the presenting phenotype. Here, the comprehensive expertise of our medical team makes a difference as we merge the patient's signs and symptoms, individual and family history, previous medical testing, and disease-related clinical and scientific information, together with the laboratory data to reach a diagnosis and determine possible therapeutic treatments or preventive measures.

Staying Current in a Rapidly Changing Field

Our medical and scientific teams participate regularly in annual conferences and professional development seminars and training on various topics related to their specialty areas to keep up with the rapidly changing field of human genetics. We work towards incorporating this knowledge into our laboratory services as part of our continuous quality improvement to ensure that clinicians get the most value from our growing portfolio of genetic services.

Sharing our Knowledge Through Professional Development & Training Events

As leading multidisciplinary experts in the field of human genetics, we assume the responsibility for imparting knowledge to other clinicians through our own annual symposia and training courses in various topics in human genetics.

Contributing our Insights and Research to Publications

Our multidisciplinary medical and scientific team contributes, continuously, to various high-profile international scientific publications in peer-reviewed journals as leading or contributing authors. To date, our team at the MGZ has submitted over 250 publications.



Research & Development

Our research & development team is actively involved in the evaluation and translation of new technologies, tests, and software applications into clinical routine testing for the MGZ's internal use as well as for external contract research.

As one of the largest genetics laboratories in Germany, the MGZ collaborates with companies, universities, as well as research organizations working in fields contributing to scientific and medical advances in the field of human genetics.

Our interdisciplinary working groups include:

- ▶ Technology Transfer & Automation
- ▶ Scientific Development
- ▶ Bioinformatics
- ▶ Software Development

Data Sharing

The MGZ collaborates in national and international consortia and various projects to further develop the field of human genetics. We publish all variants to Leiden's Open (source) Variation Database (LOVD) – an online gene-centered collection and display of DNA variants. Additionally, we are a member of the InSIGHT expert panel Variant Interpretation Committee (VIC), which meets biennially to review and discuss DNA variants that contribute to gastrointestinal tumors.



OUR INTERNATIONAL SERVICES

Your full-service provider

- ▶ Genetic Consultations for Clinicians
- ▶ Testing Strategy Recommendations
- ▶ Genetic Testing for Rare Diseases & Hereditary Cancers
- ▶ NGS and Non-NGS Diagnostics in Over 15 Clinical Areas
 - ▷ Phenotype-based Gene Panels
 - ▷ Comprehensive Testing: MGZ's DxOme®, Exome Diagnostics
 - ▷ Single Gene Analyses
 - ▷ Familial Variant Testing
 - ▷ Methylation Analyses
 - ▷ Repeat Expansion Analyses
 - ▷ Del/Dup Analyses
 - ▷ Chromosomal Microarray
 - ▷ Cytogenetics
 - ▷ Carrier Screening
 - ▷ Non-invasive Prenatal Testing (NIPT)
- ▶ Variant Interpretation by our Medical and Scientific Teams
- ▶ Professional Development Training in Medical Genetics
- ▶ Collaboration in Genetics-Related Projects
- ▶ Genetic Counseling for Patients





MGZ

ENERGOSUM
KONFERENCJA WIELKIEJ
POLSKIEJ KAPITAŁOWEJ ALIANCJI

NGS DIAGNOSTICS AT THE MGZ

Value-added diagnostic gene panels created by our experts

The MGZ offers over **250 diagnostic phenotype-based gene panels** available in over **15 clinical specialties** to serve the growing needs of clinicians.

Expertly-Designed Disease-Focused Panels

- ▶ Custom-designed by medical geneticists and scientific experts in the specific clinical disease area
- ▶ Concordant with existing, curated phenotype profiles
- ▶ Gene panels continuously updated to reflect the latest medical and scientific knowledge

Guaranteed Diagnostic Quality

- ▶ Comprehensive mutation spectrum analysis
- ▶ Specificity and sensitivity of variant detection validated for each panel, following the current NGS-based diagnostic guidelines

Comprehensive Coverage

- ▶ Complete coverage of all coding exons, in addition to manually curated regulatory/intronic regions with known pathogenic variants
- ▶ Additional coverage of specific genomic regions of interest, enabled through deep NGS analysis, resulting in more information

Targeted Approach

- ▶ Secondary findings and the incidence of variants of unknown significance (VUS) are minimized, as only genes with a known disease-association are selected for their clinical relevance and actionability.

The MGZ's DxOme®

Our custom-designed phenotype-based diagnostic gene panel with 1678 disease-relevant genes

Key Features

- ▶ **DxOme** – Three options in high-quality NGS testing for your clinical situation – from patients with a **distinct phenotype** to those with an **unspecific or complex phenotype**
- ▶ Excellent technical quality – **full technical coverage of targeted region (20-fold) of $\geq 98\%$**
- ▶ High to very **high diagnostic sensitivity**
- ▶ **Comprehensive variant interpretation** by an **experienced interdisciplinary medical and scientific team**, following the ACMG guidelines for results you can trust
- ▶ **Reliable detection of changes in gene dosage** (copy number variation (CNV), deletions/duplications) facilitated by the MGZ's special dosage bioinformatics pipeline based on NGS data for most genes
- ▶ Analysis of **single nucleotide variants (SNVs)**
- ▶ **HPO term-based expanded data interpretation**
- ▶ Gene-specific coding **pathogenic intronic and promoter variants** can be taken into account for numerous diseases
- ▶ **Technically difficult regions** can be **reliably analyzed** (e.g. GC-rich regions, low complexity regions)
- ▶ Relevant non-coding variants and panel-specific **add-on analyses where there is insufficient coverage**
- ▶ **Mitochondrial genome (mtDNA) analysis** available as an add-on test
- ▶ **Fast turnaround times:** Prenatal and urgent samples – 10 business days; Routine samples in 3-6 weeks
- ▶ **Competitive pricing** to fit in with cost-conscious institutional budgets



OUR PARTNERSHIPS & COLLABORATIONS

Benefit from our knowledge network

Due to its expertise in various genetic technologies, the MGZ – Medical Genetics Center is an interesting potential partner for a number of genetics-related projects. Research projects generally require a multidisciplinary approach, which we are able to deliver with our team of experts. This includes technology and data experts as well as clinical and scientific experts to provide the coordination and execution needed for a successful project. The MGZ – Medical Genetics Center cooperates regularly with a number of clinical and scientific institutions and is actively involved in a number of national and international projects.

- ▶ **International Society for Gastrointestinal Hereditary Tumours (InSIGHT):** The goal of this organization is to work towards improving the care of patients and families affected by a hereditary condition resulting in gastrointestinal tumours.
- ▶ **Variant Interpretation Committee (VIC):** The committee is an organization consisting of over 40 experts from multiple disciplines established to create an internationally recognized classification system for the interpretation of variants, specifically related to the mismatch repair genes for HNPCC/Lynch syndrome.
- ▶ **Leiden Open Variation Database (LOVD):** The MGZ has long been an important "submitter" for variants in the LOVD databases (LOVD 2.0 and LOVD 3.0). LOVD is an important source for the diagnostically active human geneticist in the daily interpretation of data collected in the laboratory from sequence analysis for individual patients.
- ▶ **Human Variome Project (HVP):** The project is a global initiative, founded by Richard Cotton (AUS), which collects and documents data on sequence variants with regard to their medical significance. The "Variant Effect Prediction Training Course" was established as part of the HVP. Here, the MGZ



employees are part of the organization team and actively participate in the event with lectures and workshops. Our employees are also part of the "Scientific Program Committee" of the biennial "Variant Detection" symposium.

- ▶ **European Reference Network (ERN) for Genetic Tumour Risk Syndromes (GENTURIS):** The tasks of this network include the creation of Europe-wide standards and guidelines for the recognition and care of patients with hereditary tumour syndromes, as well as the development of teaching and information materials on these diseases. Due to its expertise in this field, the MGZ was included as one of three German centers in the ERN – GENTURIS. The MGZ is particularly involved in the projects on hereditary colorectal cancer.

ACCREDITATION

Quality assured, internationally recognized



The accreditation is only valid for the scope of accreditation specified in certificate.

The MGZ – Medical Genetics Center laboratory first received accreditation in accordance with the **international standard DIN EN ISO15189** in May, 2000. This international standard requires not only the establishment and maintenance of a quality management system but also evidence of the **highest standards in genetic testing in all areas**, including pre- and post-analytics and the interpretation of test results.

The DAkkS has continually renewed the MGZ's accreditation in accordance with this international standard ever since. Additionally, in 2015, the MGZ received **accreditation for** the new technology of **Next-Generation Sequencing (NGS)**.

The DAkkS (*Deutsche Akkreditierungsstelle*, or German Accreditation Body) is the national accreditation body for the Federal Republic of Germany and acts according to EU Regulation (EC) No. 765/2008 and the Accreditation Body Act (AkkStelleG). DAkkS is a member of the international accreditation organization European co-operation for Accreditation (EA), International Accreditation Forum (IAF) and International Laboratory Accreditation Cooperation (ILAC).

The combined **ILAC-DAkkS symbol** awarded by the DAkkS indicates to accredited laboratories that their services are subject to multilateral recognition agreements and therefore enjoy **international acceptance status**.

“Setting a ‘gold standard’ in quality, safety, and reliability”

GET STARTED WITH THE MGZ TODAY

Experience the difference

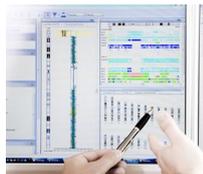
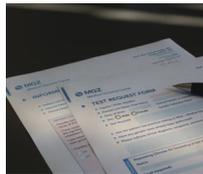
Visit our website at www.mgz-muenchen.com to explore our genetic test portfolio and learn more about our additional genetic services.

The MGZ offers:

- ▶ A comprehensive portfolio of genetic services performed at our Munich headquarters
- ▶ Easy price requests and test ordering via our website portal – Sign up today for an account to get started
- ▶ Email and telephone support for your test ordering, shipping, and billing inquiries
- ▶ Clinical support with testing strategy recommendations and case-specific consultations
- ▶ High quality diagnostics and meaningful, detailed laboratory reports to assist in clinical decision-making, therapy-relevant interventions, patient care, and familial risk assessment
- ▶ Dedicated pre- and post-analytic support to save time, to minimize processing delays for your patient samples, to discuss possible follow-up testing, and to ensure a customer-friendly experience with our services
- ▶ Multiple billing and payment options including credit card, PayPal, and wire transfers for patients as well as physicians or clinical institutes
- ▶ High volume discounts and institutional pricing available

Are you interested in our services?

Please send us an email to inquiry@mgz-muenchen.com or contact us by phone at **+49 (0) 89 / 30 90 886 - 0**. Our international support team will help get you started so that you can start realizing the benefits of working with us.





Medical Genetics Center

**CLINICAL KNOW-HOW. SCIENTIFIC EXPERTISE.
COMPREHENSIVE TESTING. ASSURED QUALITY.**

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