On our website, you will find additional information on other clinical topics as well as organizational information.

Visit us at www.mgz-muenchen.com

Prof. Elke Holinski-Feder, MD
Clinical Geneticists

Angela Abicht, MD

Stefanie Balg, MD
Brigitte Schönfeld
Teresa Neuhann, MD
Kerstin Becker, MD
Daniela González Fassrainer
Verena Steinke-Lange, MD
Yvonne Müller-Koch, MD
Anne Behnecke, MD
Clinical Geneticists
Silja Gnann, MD
Internist
Clinical Geneticist
Isabel Diebold, MD
Pediatrician

MGZ – Medical Genetics Center

Prof. Elke Holinski-Feder, MD
Angela Abicht, MD
Clinical Geneticists, MVZ

Bayerstrasse 3 - 5 | D-80335 Munich
Phone +49 (0)89 / 30 90 886 - 0 | Fax +49 (0)89 / 30 90 886 - 66
inquiry@mgz-muenchen.com | www.mgz-muenchen.com

ilac-MRA DAkkS Deutsche Akkreditierungsstelle D-ML-13242-01-00

Version: AI01E_ENG_0005_02.6/2018

MGZ
Medical Genetics Center

Clinician Information

CARRIER SCREENING

WHAT IS CARRIER SCREENING?

The aim of carrier screening is to identify individuals or couples with an increased risk of having children affected by genetic diseases.

It is assumed that each person in the general population is a heterozygous carrier of 2 to 3 recessive mutations known to cause severe childhood diseases when present in the maternal and paternal copies of a gene. Although individual genetic diseases are rare in general populations, collectively they account for ~20 % of infant mortality and ~10 % of pediatric hospitalizations.[1] The diagnosis of some rare genetic diseases also has therapeutic consequences, so that in these cases making a quick, early diagnosis is extremely important.

Unlike traditional ethnic screening, carrier screening is offered to all individuals regardless of their ethnicity (non-directive, pan-ethnic carrier screening). The American College of Obstetricians and Gynecologists (ACOG) recently recognized pan-ethnic carrier screening as an acceptable strategy for pre-pregnancy and prenatal carrier screening (ACOG Committee Opinion No. 690 2017).

Like any genetic analysis, carrier screening does not have 100 % sensitivity. An inconspicuous result means a significant risk reduction. Patients should be advised on the residual risk for each test result.

WHO SHOULD PURSUE CARRIER SCREENING?

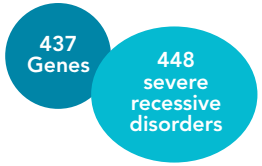
Carrier screening is recommended for the following:

- Consanguineous couples
- Individuals with unidentified recessive genetic diseases in the family
- Couples undergoing IVF/fertility treatment
- Early pregnancy
- Family planning, even when there are no known genetic diseases in the family

WHAT DOES MGZ OFFER IN TERMS OF CARRIER SCREENING?

MGZ offers carrier screening for the following:

- **Fragile X syndrome**
- **Spinal muscular atrophy (SMA)**
- **Thalassemias** and **hemoglobinopathies**
- **448 severe recessive diseases of childhood** caused by pathogenic variants in 437 genes ("Kingsmore Panel"). Cystic fibrosis (CF) is included in this testing. The genes of this next-generation sequencing (NGS) panel were selected and evaluated by experts in accordance with the recommendations of the American College of Medical Genetics and Genomics (ACMG). Criteria were e.g., high penetrance, sufficient published data regarding clinical sensitivity / specificity, functional studies, pathogenic variants found in more than one family, etc.[1]



WHAT ARE THE BENEFITS OF CARRIER SCREENING AT MGZ?

In combination with genetic counseling, carrier screening can assist in making individual decisions in family planning.

- If it appears that a person is a carrier of a specific condition, his/her reproductive partner should be offered screening to provide accurate genetic counseling regarding the risk of having an affected child.
- Identification of carrier status for a couple enables individuals to consider the full range of available reproductive options (e.g. donor gametes, pre-implantation genetic diagnosis, prenatal diagnosis).
- Carrier status for certain diseases may raise awareness of maternal obstetric complications and improve pregnancy management.
- If a specific familial mutation is known, this may enable specific and rapid postnatal diagnosis for newborns, which may have consequences for better treatment options.

HOW TO ORDER CARRIER SCREENING AT MGZ

- **Specimen (preferably from both partners):**
  - 2 - 4 mL EDTA blood
  - From 1,5 µg DNA (preferably > 3 µg)
- **Turnaround time**
  - 3 - 6 weeks
- **Price Inquiries & Test Ordering**

  To request price information, send an e-mail to inquiry@mgz-muenchen.com
  For detailed information regarding our services and areas of clinical expertise, please visit our website: www.mgz-muenchen.com

Literature:
[1] Bell CJ et al. Carrier testing for severe childhood recessive diseases by next-generation sequencing. Sci Transl Med. 2011; 3:65ra4.
[2] Kauffman TL et al. Design of a randomized controlled trial for genomic carrier screening in healthy patients seeking preconception genetic testing. Contemporary Clinical Trials 2017; 53:100–105
[3] Ghiossi CE et al. Clinical Utility of Expanded Carrier Screening: Reproductive Behaviors of At-Risk Couples. J Genet Counsel 2017; https://doi.org/10.1007/s10897-017-0160-1
[4] Lazarin EA and Haque IS. Expanded carrier screening: A review of early implementation and literature. Semin Perinatol. 2016; 40:29-34.