Order form for molecular genetic testing
Zentrum Medizinische Genetik Würzburg, Biozentrum, Am Hubland, 97074 Würzburg

Patient details (use label):

- Male
- Female

Given name

Name

Date of birth

Cost coverage declaration:

- Invoice to patient (give details and address on reverse page)
- Invoice to referring institution
- Invoice by special agreement with Zentrum Medizinische Genetik Würzburg (to be obtained prior to testing)
- Invoice to patient (give details and address on reverse page)

Relevant clinical patient and/or family data (please enclose reports, if available):

- Clinical diagnostics
- Predictive diagnostics
- Known Index patient / known mutation
  - Gene: ____________________
  - Mutation: ____________________

Test order (please tick boxes)

- Neuro-muscular disorders
  - DMD/BMD (del/dup), (MLPA)
  - DMD/BMD (point mutations)
  - Emery-Dreifuss muscular dystrophy (aut. dom.)
  - Emery-Dreifuss muscular dystrophy (X-linked)
  - Facio-scapulo-humeral MD 1 (FSHD1, D4Z4)
  - Facio-scapulo-humeral MD 2 (FSHD2, SMCHD1)
  - Muscle hypertrophy (Myostatin)*
  - Myofibrillar myopathy (MFM)
  - Myofibrillar myopathy (MFM)
  - Myotubular myopathies
    - B1N1 (autosomal recessive)
    - DNM2 (autosomal dominant)
    - MMT1 (X-linked)
  - Myotonic dystrophies
    - DM1 (Curschmann-Steinert)
    - DM2 (Proximal myotonic myopathy)

- Structural myopathies – Malignant Hyperthermia
  - Central core disease (RYR1)
  - Malignant hyperthermia (RYR1)
  - Multi minicore disease (SEPN1/RYR1)
  - Nemaline myopathy (ACTA1)
  - Limb girdle muscular dystrophies
    - LGMD 1B (Lamin A/C)
    - LGMD 1C (Cavelin3)
    - LGMD 2A (Calpain 3)
    - LGMD 2B (Dysferlin)
    - LGMD 2D (SGCA)
    - LGMD 2E (SGCB)
    - LGMD 2G (Telethonin)*
    - LGMD 3 (FMRP)
    - LGMD 2I (Mitochondrial, Titin)
    - LGMD 2L (anos)

- Structural myopathies – Malignant Hyperthermia

- Craniolensostoses
  - Apert syndrome (FGFR2, hot spot)
  - Carpenter syndrome (RBSB2)*
  - Crouzon syndrome (FGFR2, hot spot)
  - FGFR2 (whole gene)
  - Coronal craniosynostosis (TGF12*)
  - LADD syndrome (FGF10*)
  - Muenke syndrome (FGFR3)
  - Pfeiffer syndrome (FGFR2 and FGFR1)
  - Saethre-Chotzen syndrome (TWIST)
  - FGFR3 associated skeletal dysplasias
    - Achondroplasia
    - Hot spot, whole gene
    - FGFR3 (hot spot)
    - Hypochondroplasia
    - Hot spot, whole gene
    - Thanatophoric Dysplasia

- Limb malformations
  - Split hand/foot* SHFM3*, SHFLD3*
  - TAR-Syndrome (Del 1q21.1, RBM8A*)
  - Others
    - ADPKD
    - PKD1, PKD2
    - Akapturina (HGD)*
    - CM1/HNPP (PMP22/MLPA)*
    - Hypophosphatasia (ALPL)
    - Kalmann syndrome (FGFR1, whole gene)
    - Short stature (SHOX)
    - Lipodyplastic (LMNB2*)
    - Marfan syndrome (FBN1)
    - Micro-deletion screening (MLPA)
    - Neurofibromatosis (NF1)
    - Pyruvate kinase deficiency (PKLR)

- If you want to order more myopathy genes, please use the order form for Myopathy Panel

MATERIAL: 5-10 ml EDTA- blood. Please label tubes clearly, package in shatter-resistant packaging; ship at ambient temperature as soon as possible within one week.

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Sample taken (date): ____________________ by: ____________________

Doctor’s name (please print) ____________________

(Doctor’s stamp) ____________________

According to the German gene testing act written patient’s consent is required for every genetic test (see page 2)
The German gene testing act (GenDG) requires written informed consent to be obtained from every patient prior to genetic testing.

Please read the following carefully, make sure all your questions were answered and tick boxes as appropriate.

<table>
<thead>
<tr>
<th>My doctor has informed me about the following diagnosis / disorder / syndrome:</th>
<th>□ yes</th>
<th>□ no</th>
</tr>
</thead>
<tbody>
<tr>
<td>its genetic basis, options for prevention and treatment and about the scope and aims of the planned genetic test, its predictive value and its limits. I have been informed about the risk of the required blood / tissue sampling. All my questions have been answered to my satisfaction.</td>
<td>□ yes</td>
<td>□ no</td>
</tr>
</tbody>
</table>

I consent that the results of the genetic test(s) are also sent to my other medical professionals, specifically to:

Dr.: ........................................................................................................................................................................

The application of such screening tests can result in incidental findings, which are not associated with the above named disease. I wish to be informed of any such incidental findings.

By German law, surplus genetic material (blood, DNA sample) must be destroyed after the completion of the genetic test. However, with my consent it may be stored and used for subsequent additional tests (if required) and/or as control for later testing of family members and relatives.

I consent to storage and subsequent use of my genetic material and/or the genetic material of my child for the above purposes.

Internal quality control is an important tool to guarantee the accuracy and reliability of genetic testing methods. For this purpose, genetic material from patients with rare genetic variants is an indispensable control material.

I consent to my DNA and/or the DNA of my child to be stored and used for internal quality control in the laboratory. Before such use, my sample and/or the sample of my child will be anonymised.

Genetic material from patients is also important for studying biological mechanisms which contribute to the development of hereditary diseases.

I consent to my DNA and/or the DNA of my child to be stored and used for potential disease studies in the laboratory. I consent to be re-contacted before such use.

The German gene testing act requires genetic results to be stored for 10 years and then destroyed. With patient’s consent they may be stored for longer. Often, genetic results are required for counselling of children and relatives even after 10 years’ time.

I consent to storage of my genetic results and/or the results of my child beyond the legal time-span and its use for my family only.

As required the results may be used for the counselling / analysis of my relatives.

Genetic data will be deposited in a database at the Institut für Humangenetik. Selected data will be anonymised and only used for the purpose of quality control and data comparison.

I have been informed that I can withdraw my consent at any time without giving reason and without incurring any penalty. I have further been informed that I have the right not to know about my genetic test results and to terminate the testing procedure at any time. I can request my genetic material and/or the genetic material of my child and my genetic results and reports and/or the genetic results and reports of my child to be destroyed before result reporting, if I have changed my mind.

With my signature I consent to the genetic test(s) indicated above and the sampling of blood or tissue for this purpose.

__________________________  ________________________
City, date                  Signature of the patient or his/her legal representative