Order form for myopathy panel sequencing

Zentrum Medizinische Genetik Würzburg, Biozentrum, Am Hubland, 97074 Würzburg

Patient details (use label):

☐ male ☐ female

Name

Given name Date of birth

Relevant clinical patient and/or family data:

Information to anamnesis and other remarks:
(please enclose report, if available)

Subpanels (please tick boxes)

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

Cost coverage declaration (mandatory to fill in):

☐ **A signed agreement to cost coverage is enclosed**

Please send us a cost quote for an:

☐ **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)**

The invoicing process will be handled on our behalf by Ärztliche Verrechnungsstelle Büdingen e.V.

Information about the myopathy panel:

The technology „Next generation sequencing" (NGS) allows the parallel analysis of numerous genes in a single approach permitting panel diagnostics. We have designed and validated a panel of **65 genes** which are known to be responsible for the most common hereditary forms of muscular diseases.

Unless otherwise ordered, we will analyze all genes on the panel. However, if you would like the analysis to be restricted to a sub-group of 'core genes', please tick the boxes to specify the subpanel.

The analysis will be performed by Illumina Nextera Capture and MiSeq technologies. Together with the report you will receive an overview of the analyzed genes and the achieved coverage.

The NGS technology covers only mutations, which are detectable by DNA sequencing. Other types of mutations like repeat expansions (e.g. in the myotonic dystrophies), large deletions and duplications (SMN1 or dystrophin gene deletions) and methylation deficiencies cannot be analyzed. Furthermore, the complex genetic alterations underlying FSHD cannot be analyzed.

We are happy to answer further questions.

Contacts:
Dr. Gerhard Meng, tel: +49-931-31-84064 (cost information)
Dr. Wolfram Kress, tel: +49-931-31-84062 (clinical information)
Konstantinos Kolokotronis, tel: +49-931-31-82187 (clinical information)
PD Dr. Simone Rost, tel: +49-931-31-84095 (technical information)

➢ Please note our order form for single gene analysis (e.g. DM1, DM2, FSHD1, SMA, etc.) on our homepage:
http://www.humgen.biozentrum.uni-wuerzburg.de/patientenversorgung/formulare/

☐ ***Muscular dystrophies (MD)***
Duchenne and Becker MD (DMD, BMD)
Emery-Dreifuss MD (aut. dominant, LMNA, X-linked , EMD)
Facioscapulohumeral muscular MD, type 2 (FSHD2, SMCHD1)
Rigid Spine muscular dystrophy (SEPN1),
Bethlem-/Ullrich myopathy (COL6A1, COL6A2, COL6A3)

☐ ***Structural myopathies***
Nemaline myopathy (ACTA1, TNNT1, TPM2, TPM3)
Myotubular (centronuclear) myopathies (BIN1, DNM2, MTM1, RYR1)
Central-core-, multi-mini-core-disease (ACTA1, RYR1, SEPN1)

☐ ***Myofibrillar and distal myopathies***
MFM (BAG3, CRYAB, DES, DNAJB6, FHL1, FLNC, MYOT, ZASP)
Distal myopathies (DES, DNAJB6, FLNC, KLHL9, MYH7, TIA1)

☐ ***Limb girdle muscular dystrophies with high CK value (LGMD-A)***
LGMD 2A (CAPN 3), LGMD 2B (DYSF), LGMD 2C (SGCG),
LGMD 2D (SGCA), LGMD 2E (SGCB), LGMD 2F (SGCD),
LGMD 2G (TCAP), LGMD 2i (FKRP), LGMD 2L (ANO5),
LGMD 1A (MYOT), LGMD 1C (CAV3), GSD2 (GAA), GSD5 (PYGM)

☐ ***Congenital & further limb girdle muscular dystrophies (LGMD-B)***
LGMD 2H (TRIM32), LGMD 2i (FKRP), LGMD 2K (POMT1),
LGMD 2M (FKTN), LGMD 2N (POMT2), LGMD 2O (POMGNT1),
LGMD 2P (DAG1), LGMD 2S (TRAPPC11), LGMD 2T (GMPPB),
LGMD 2U (ISPD), LGMD 1B (LMNA), LGMD 1F (TNPO3)

☐ ***Complete myopathy panel***

Samples taken (date): _____ by: _____

_____
Physicians name (please print)

_____ _____
Date Physicians signature (Physicians stamp)

According to the German gene testing act written patient's consent is required for every genetic test (see page 2)

Consent for genetic testing using panel diagnostics

Patient details (use label):

Name _____ Given name _____

Date of birth _____ ☐ male ☐ female

Street _____

Post code _____ City _____

Please return to:

Zentrum Med. Genetik Würzburg
Biozentrum, Am Hubland
97074 Würzburg

Praxis für Humangenetik
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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, and tick **each** box, as appropriate*

I hereby consent to the analysis of my DNA or the DNA of my child by panel diagnostics for clarification of a putative **hereditary muscle disease.**

I have been informed about 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 further informed about the risk of the required blood / tissue sampling. All my questions have been answered to my satisfaction.

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.	☐ yes ☐ no
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.	☐ yes ☐ no
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.	☐ yes ☐ no
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.	☐ yes ☐ no
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.	☐ yes ☐ no
As required the results may be used for the counselling / analysis of my relatives.	☐ yes ☐ no

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

Source: http://www.humgen.biozentrum.uni-wuerzburg.de/ patientenversorgung/formulare/

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