

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 / prenatal / carrier diagnostics
- Known Index patient / known mutation: yes no

Gene: _____ Mutation: _____

Test order (please tick boxes)

Please note the separate test orders for panel diagnostics on our homepage: gene panels for Fanconi and Tumour disease, Myopathy, Craniosynostoses, CMTs, SPGs, Cardiomyopathies and Hereditary Hearing Loss.

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

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)*
- Myoadenylate desaminase def. (MADD)(Hot spots)
- Oculo-pharyngeal muscular dystrophy (OPMD)
- Rigid Spine muscular dystrophy (SEPN1)
- Spinal muscular atrophies (5q-associated)
- Spino-bulbar muscular atrophy (Kennedy)

Myotubular myopathies

- BIN1 (autosomal recessive)
- DNM2 (autosomal dominant)
- MTM1 (X-linked)

Myofibrillar myopathies (MFM)

- BAG3
- CRYAB
- DES
- DNAJB6*
- FHL1
- FLNC
- MYOT
- TTN (Hot spots)*
- LDB3 (ZASP)

Distal Myopathies

- DES
- DNAJB6*
- FLNC
- MYH7

Myotonic dystrophies

- DM1 (Curschmann-Steinert)
- DM2 (Proximal myotonic myopathy)

Structural myopathies – Malignant Hyperth.

- Central core disease (RYR1)
- Malignant hyperthermia (RYR1/CACNA1S*)
- Multi minicore disease (SEPN1/RYR1)
- Nemaline myopathy (ACTA1)

Limb girdle muscular dystrophies

- Type EDMD (LMNA)
- Type RMD (CAV3)
- Type LGMD R1 (CAPN3)
- Type LGMD R2 (DYSF)
- Type LGMD R3 (SGCA)
- Type LGMD R4 (SGCB)
- Type LGMD R7 (TCAP)*
- Type LGMD R9 (FKRP)
- Type LGMD R10 (TTN, Hot spots)
- Type LGMD R12 (ANO5)

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

Neuro-degenerative disorders

- Chorea Huntington (HTT)

Coagulation disorders

- Hemophilia A (F8)
- Hemophilia B (F9)
- Hereditary angioedema type I & II (Serping1)

Hereditary hearing loss

- CX26 (GJB2)*, CX30 (GJB6)*
- STRC*

Craniosynostoses

The order form for the Craniosynostoses can be found on our homepage

FGFR3 associated skeletal dysplasias

- Achondroplasia (FGFR3)
- Hypochondroplasia (FGFR3)
- Thanatophoric Dysplasia (FGFR3)
- Split hand/foot* SHFM3*, SHFLD3*
- TAR-Syndrome (Del 1q21.1; RBM8A)*

Others

- Alkaptonuria (HGD)*
- CMT 1A/HNPP, (PMP22, MLPA)*
- Hypophosphatasia (ALPL)
- Short stature (SHOX)
- Lipodystrophy (LMNB2)*
- Marfan syndrome (FBN1)
- Micro-deletion screening (MLPA)
- Neurofibromatosis (NF1)
- Polycystic kidney disease (PKD1, PKD2)
- Pyruvate kinase deficiency (PKLR)

* not accredited

Sample taken (date): _____ by: _____

Doctor's name (please print) _____

Doctor's signature _____

(Doctor's stamp)

According to the German gene testing act written patient's consent is required for every genetic test
<https://www.biozentrum.uni-wuerzburg.de/humangenetik/patientenversorgung/forms-en/>