

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)

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:
<https://www.biozentrum.uni-wuerzburg.de/humangenetik/patientenversorgung/formulare/>

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

Distal Myopathies

- ☐ DES ☐ FLNC
☐ DNAJB6* ☐ 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)

Limb malformations

- ☐ 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