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:
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
- ☐ 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)

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
https://www.biozentrum.uni-wuerzburg.de/humangenetik/patientenversorgung/forms-en/