

Order form for molecular genetic testing

University hospital Würzburg, Institute for Clinical Genetics and Genomic Medicine, 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 Institute for Clinical Genetics and Genomic Medicine (to be obtained prior to testing)

Tel.: 0931-201-29931, Fax: 0931-201-6029931
E-Mail: Diagnostik_KGGM@ukw.de

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 the homepage:
<https://www.ukw.de/klinische-genetik-und-genommedizin/diagnostik/>

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 Hypertherm.

- 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)
 Pyruvate kinase deficiency (PKLR)

* not accredited

Sample taken (date): _____ by: _____

Doctor's name (please print) _____

(Doctor's stamp)

Doctor's signature _____

According to the German gene testing act written patient's consent is required for every genetic test.