Consent for genetic testing

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
PD Dr. med. Erdmute Kunstmann
Tel: 0931-3184435, Fax: 0931-45265859
E-Mail: kunstmann@biozentrum.uni-wuerzburg.de

Institut für Humangenetik, DNA-Labor
Tel: 0931-3184064, Fax: 0931-3184069
E-Mail: gmeng@biozentrum.uni-wuerzburg.de

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, make sure all your questions were answered and tick boxes as appropriate.

My doctor has informed me about the following diagnosis / disorder / syndrome : .... 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 informed about the risk of the required blood / tissue sampling. All my questions have been answered to my satisfaction.	☐ yes
I consent that the results of the genetic test(s) are also sent to my other medical professionals, specifically to Dr.:	☐ yes ☐ no
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 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 being stored and used for internal quality control in the laboratory. Before such use, my sample 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 being stored and used for disease studies in the laboratory. Before such use, my sample will be anonymised.	☐ yes ☐ no
The German gene testing act requires genetic data to be stored for 10 years and then destroyed. With patient's consent they may be stored for longer. Often, genetic data are required for counselling of children and relatives even after 10 years time. I consent to storage of my genetic data beyond the legal time-span and its use for my family only.	☐ yes ☐ no

Genetic data will be deposited in a database at the Institut für Humangenetik. All data will 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 having disadvantage. 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 my genetic data 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

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 disease, Tumour, Myopathy, Craniosynostoses** 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 deficiency (MADA)
- ☐ Myopathy-Brody (ATP2A1)*
- ☐ Oculo-pharyngeal muscular dystrophy (OPMD)
- ☐ Rigid Spine muscular dystrophy (SEPN1)
- ☐ Spinal muscular atrophies (type I, II and III)
- ☐ Spino-bulbar muscular atrophy (Kennedy)

Myotubular myopathies

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

Myofibrillar myopathies (MFM)

- ☐ BAG3
- ☐ CRYAB
- ☐ Desmin
- ☐ DNAJB6*
- ☐ FHL1
- ☐ FLNC
- ☐ MYOT (Hot spots)
- ☐ TTN (Hot spots)*
- ☐ ZASP (Hot spots)

Distal Myopathies

- ☐ Desmin
- ☐ DNAJB6*
- ☐ FLNC
- ☐ MYH7

Myotonic dystrophies

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

Structural myopathies – Malignant Hyperth.

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

Limb girdle muscular dystrophies

- ☐ LGMD 1B (Lamin A/C)
- ☐ LGMD 1C (Caveolin3)
- ☐ LGMD 2A (Calpain 3)
- ☐ LGMD 2B (Dysferlin)
- ☐ LGMD 2D (SGCA)
- ☐ LGMD 2E (SGCB)
- ☐ LGMD 2G (Telethonin)*
- ☐ LGMD 2i (FKRP)
- ☐ LGMD 2J (tibial MD, Titin)
- ☐ LGMD 2L (ANO5)

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

Neuro-degenerative disorders

- ☐ Chorea Huntington (HTT)
- ☐ Pontocerebellar hypoplasia type 2 (TSEN54)

Coagulation disorders

- ☐ Hemophilia A (factor VIII)
- ☐ Hemophilia B (factor IX)
- ☐ Hereditary angioedema type I & II (C1 inhibitor)
- ☐ von Willebrand disease type 2N (Normandy)

Hereditary hearing loss

- ☐ CX26 (GJB2)*,
- ☐ CX31 (GJB6)*
- ☐ CX31 (GJB3)*,
- ☐ STRC*

* not accredited

Craniosynostoses

- ☐ Apert syndrome (FGFR2, hot spot)
- ☐ Carpenter syndrome (RAB23)*
- ☐ Crouzon syndrome (FGFR2, hot spot)
- ☐ FGFR2 (whole gene)
- ☐ Foramina parietalia ☐ ALX4, ☐ MSX2,
- ☐ Coronal craniosynostosis (TCF12)*
- ☐ LADD syndrome (FGF10)*
- ☐ Muenke syndrome (FGFR3)
- ☐ Pfeiffer syndrome (FGFR2 and FGFR1)
- ☐ Saethre-Chotzen syndrome (TWIST)

FGFR3 associated skeletal dysplasias

- ☐ Achondroplasia ☐ Hot spot, ☐ whole gene
- ☐ FGFR3 (hot spot)
- ☐ Hypochondroplasia ☐ Hot spot, ☐ whole
- ☐ Thanatophoric Dysplasia

Limb malformations

- ☐ Split hand/foot* ☐ SHFM3*, ☐ SHFLD3*
- ☐ TAR-Syndrome (Del 1q21.1; RBM8A)*

Others

- ☐ ADPKD ☐ PKD1, ☐ PKD2
- ☐ Alkaptonuria (HGD)*
- ☐ CMT 1/HNPP (PMP22)(MLPA)*
- ☐ Hypophosphatasia (ALPL)
- ☐ Kallmann syndrome (FGFR1, whole gene)
- ☐ Short stature (SHOX)
- ☐ Lipodystrophy (LMNB2)*
- ☐ Marfan syndrome (FBN1)
- ☐ Micro-deletion screening (MLPA)
- ☐ Neurofibromatosis (NF1)
- ☐ Pyruvate kinase deficiency (PKLR)

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 (see overleaf)