UNIKLINIK KÖLN | Institut für Humangenetik

MVZ Medizinisches Versorgungszentrum der Uniklinik Köln am Neumarkt

PATIENT INFORMATION (use sticker if available)

Last name _____

First name(s) _____

Date of birth _____

Address _____

_____

female ☐ **male** ☐

Ethnic background _____
(may be important in recessive conditions)

Institute of Human Genetics
University Hospital of Cologne
Kerpener Str. 34
50931 Cologne
Germany

Phone +49/221/478-86811, Fax +49/221/478-86812
www.uk-koeln.de\humangenetik

Billing

Test will be paid by ☐ referring facility ☐ patient

Please note that international requests must be accompanied by a confirmation of payment. Please contact us for details.

Request for molecular genetic testing

See page 2 for available tests

Reason for testing:

Please provide pedigree / clinical findings / details on pregnancy (week), previous genetic tests perfomed, if appropriate.

Informed consent form for genetic testing ("DNA analysis")

According to the German Genetic Diagnostics Act (www.bvdh.de/newsdownload/40/Gesetzblatt_GenDG_BGBL04082009.pdf)

1.) I herewith consent that genetic testing will performed on a blood/biological sample derived from
☐ **me** ☐ **my child** ☐ **the person under my legal guardianship**
I have received full information from my physician concerning the suspected diagnosis of

_____ ,
its genetic basis and the possible interpretations and limitations of the diagnostic testing.

2.) I herewith consent that the genetic test results will not be destroyed after 10 years as laid down in German statutory provisions but will be retained so that they will be available to me and/or members of my family.

3.) I herewith consent that the test results will be stored in hard copy and as electronic files in accordance with legal provisions and that they will be used without disclosing personal data (i.e. in pseudonymized form) for scientific or quality management purposes.

4.) I herewith consent that, after the requested testing has been completed, the Institute of Human Genetics, University Hospital of Cologne, may use the remaining sample material without disclosing personal data (i.e. in pseudomomized form) for quality management, teaching and/or scientific purposes.

5.) Results of the above stated genetic testing may be disclosed to the following attending physician(s):

_____

— Please delete as appropriate —

I am free to withdraw any of the above statements in writing without giving any reasons. Such withdrawal will involve no loss of benefits for me.

Referring physician

_____
Signature Place, Date

_____
Name

_____
Institution

_____

_____
Phone, Fax

Patient/legal guardian

_____
Signature Place, Date

_____
Name

_____
Address

_____

_____
Phone

►Please give a copy of the completed and signed consent form to the patient◄

Institute of Human Genetics, University Hospital of Cologne, Kerpener Str. 34, 50931 Cologne, Germany, Phone +49/221/478-86811

Molecular genetic request form

Neuromuscular Disorders (Contact: brunhilde.wirth@uk-koeln.de, nadine.plume@uk-koeln.de, jutta.becker@uk-koeln.de, raoul.heller@uk-koeln.de)

Spinal muscular atrophy type I-IV (SMA); recessive

- ☐ *SMN1* deletion test (MLPA)
- ☐ *SMN1* carrier test (MLPA)
- ☐ *SMN1* point mutation analysis (sequencing) (upon inquiry)
- ☐ *SMN2* (MLPA)

X-linked SMA; X-recessive

- ☐ *UBA1* (sequencing)

Amyotrophic lateral sclerosis (ALS); familial

- ☐ *SOD1* (sequencing)
- ☐ *ALS2* (sequencing)
- ☐ *VAPB* (sequencing

Spinal muscular atrophy with respiratory distress type 1 (SMARD1), diaphragmatic SMA (DSMA1); recessive

- ☐ *IGHMBP2* (sequencing)

Charcot-Marie-Tooth 2C (HMSN IIc), scapuloperoneal SMA, distal benign SMA with contractures; dominant

- ☐ *TRPV4* (sequencing)

Arthrogryposis (AMC), distal (DA1, DA2A, DA2B, DA7); dominant

- ☐ *TPM2* (sequencing)
- ☐ *TNNI2* (sequencing)
- ☐ *TNNT3* (sequencing)
- ☐ *MYH3* (sequencing)
- ☐ *MYH8* (sequencing)
- ☐ *MYBPC1* (sequencing)

Fetal akinesia deformation sequence (FADS), Pena-Shokeir; recessive

- ☐ *RAPSN* (sequencing)
- ☐ *CHRNG* (sequencing)
- ☐ other (upon inquiry)

Congenital myopathy (fiber-type disproportion); dominant

- ☐ *ACTA1* (sequencing)
- ☐ *SEPN1* (sequencing)
- ☐ other (upon inquiry)

Pontocerebellar hypoplasia (PCH 2 and 4); recessive

- ☐ *TSEN54* (sequencing)
- ☐ *TSEN2* (sequencing)
- ☐ *TSEN34* (sequencing)
- ☐ other (upon inquiry)

Skeletal Disorders (Contact: christian.netzer@uk-koeln.de, lutz.garbes@uk-koeln.de, jutta.becker@uk-koeln.de)

Osteogenesis imperfecta (OI) type I – IV; dominant

- ☐ *COL1A1* (sequencing, MLPA)
- ☐ *COL1A2* (sequencing, MLPA)

Osteogenesis imperfecta (OI) type IIB und VII; recessive

- ☐ *CRTAP* (sequencing)
- ☐ *FKBP10* (sequencing)
- ☐ *LEPRE1* (sequencing)
- ☐ *PPIB* (sequencing)
- ☐ *SP7* (sequencing)
- ☐ *SERPINH1* (sequencing)
- ☐ *SERPINF1* (sequencing)

Kidney Disorders (Contact: bodo.beck@uk-koeln.de, nadine.plume@uk-koeln.de)

Primary hyperoxaluria type 1 (PH I); recessive

- ☐ *AGXT* (sequencing, MLPA)

Primary hyperoxaluria type 2 (PH II); recessive

- ☐ *GRHPR* (sequencing, MLPA)

Primary hyperoxaluria type 3 (PH III); recessive

- ☐ *DHDPSL* (sequencing)

Nephrotic syndrome; recessive

- ☐ *NPHS1* (sequencing)
- ☐ *NPHS2* (sequencing)
- ☐ *WT1* (sequencing)

Medullary cystic kidney disease (MCKD)/ Urinary tract malformations; dominant

- ☐ *UMOD* (sequencing)
- ☐ *HNF1β* (sequencing, MLPA)
- ☐ *REN* (sequencing)

Renal-tubular dysgenesis (RTD); recessive

- ☐ *ACE* (sequencing)
- ☐ *AGT* (sequencing)
- ☐ *AGTR2* (sequencing)
- ☐ *REN* (sequencing)

Kabuki Syndrome (Contact: bwollnik@uni-koeln.de, jutta.becker@uk-koeln.de)

- ☐ *MLL2* (sequencing)

Craniofacial Malformation Syndromes (Contact: bwollnik@uni-koeln.de, jutta.becker@uk-koeln.de)

Syndromic craniosynostoses; dominant (incl. Alpert, Pfeiffer, Crouzon, Saethre-Chotzen, Muenke syndromes)

- ☐ *FGFR1* (sequencing, hot spots)
- ☐ *FGFR2* (sequencing, hot spots)
- ☐ *FGFR3* (sequencing, hot spots)
- ☐ *TWIST* (sequencing, MLPA)

LADD syndrome, ALSG syndrome; dominant

- ☐ *FGF10* (sequencing, MLPA)
- ☐ *FGFR2* (sequencing, TK domain)
- ☐ *FGFR3* (sequencing, TK domain)

Hearing Disorders (Contact: christian.netzer@uk-koeln.de, jutta.becker@uk-koeln.de)

Autosomal recessive/digenic hearing loss

- ☐ *GJB2 (Cx 26)* (sequencing)
- ☐ *GJB6* (Cx 30) (PCR of junction fragment)

Pendred syndrome/DFNB4; recessive

- ☐ *SLC26A4* (sequencing)

Multisystem Disorders (Contact: christian.netzer@uk-koeln.de, lutz.garbes@uk-koeln.de, jutta.becker@uk-koeln.de)

Cystic fibrosis; recessive

- ☐ *CFTR* (hot spots, OLA)
- ☐ *CFTR* (sequencing)
- ☐ *CFTR* (sequencing, MLPA)

Sample and shipping requirements

5-10 ml EDTA blood / ≥10 ml amniotic fluid / chorionic villi / ≥500 ng DNA;
1-2 ml EDTA blood acceptable for newborns and infants (please contact us).
Please contact us before submitting samples for prenatal diagnosis/during pregnancy.

Ship samples at room temperature. Please make sure that samples are correctly **labelled (name & dob)**!
Testing will only be performed if samples are accompanied by a completed and signed informed consent form (s. page 1).