



**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 performed, 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 be 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 pseudonymized 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 \_\_\_\_\_

## Molecular genetic request form

### NEUROMUSCULAR DISORDERS (contact: brunhilde.wirth@uk-koeln.de, nadine.reintjes@uk-koeln.de, lutz.garbes@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)

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

- IGHMBP2 (sequencing, MLPA)

#### *Amyotrophic lateral sclerosis (ALS); familial*

- SOD1 (sequencing)
- ALS2 (sequencing)
- VAPB (sequencing)

#### *Pontocerebellar hypoplasia (PCH 2 and 4); recessive*

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

#### *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)
- DOK7 (sequencing)
- other (upon inquiry)

#### *Congenital myopathy (fiber-type disproportion); dominant*

- ACTA1 (sequencing)
- SEPN1 (sequencing)
- MUSK (sequencing)
- other (upon inquiry)

### SKELETAL DISORDERS (contact: christian.netzer@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, VII, VI, X; recessive*

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

### KIDNEY DISORDERS (contact: bodo.beck@uk-koeln.de, nadine.reintjes@uk-koeln.de)

#### *Nephrotic syndrome; recessive*

- NPHS1 (sequencing)
- NPHS2 (sequencing)
- PLCE1 (NPHS3) (sequencing)
- LAMB2 (sequencing)
- SMARCAL1 (sequencing)

#### *Nephrotic syndrome; dominant*

- WT1 (sequencing)
- LMX1B (Nail-patella syndrome) (sequencing)

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

- UMOD (MCKD2) (sequencing)
- HNF1B (sequencing, MLPA)
- REN (sequencing)

#### *Renal-tubular dysgenesis (RTD); recessive*

- ACE (sequencing)
- AGT (sequencing)
- AGTR1 (sequencing)
- REN (sequencing)

#### *Urolithiasis/Nephrocalcinosis*

#### *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*

- DHAPSL (sequencing)

#### *Dent disease; X-recessive*

- CLCN5 (sequencing)

### KABUKI SYNDROME (contact: bwollnik@uni-koeln.de, jutta.becker@uk-koeln.de)

#### *Kabuki syndrome; dominant*

- MLL2 (sequencing, MLPA)

## Molecular genetic request form

### CRANIOFACIAL MALFORMATION SYNDROMES (contact: bwollnik@uni-koeln.de, lutz.garbes@uk-koeln.de)

#### *Syndromic craniosynostoses; dominant*

(incl. Alpert, Pfeiffer, Crouzon, Saethre-Chotzen, Muenke syndromes)

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

#### *LADD syndrome, ALSG syndrome; dominant*

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

#### *Hypochondroplasia, Achondroplasia; dominant*

- FGFR3* (sequencing, hot spots)

### HEARING DISORDERS (contact: christian.netzer@uk-koeln.de, lutz.garbes@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, jutta.becker@uk-koeln.de, bodo.beck@uk-koeln.de, nadine.reintjes@uk-koeln.de)

#### *Cystic fibrosis; recessive*

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

#### *MODY diabetes (Maturity Onset Diabetes of the Young); dominant*

- HNF1A* (sequencing, MLPA)       *HNF4A* (sequencing, MLPA)  
 *HNF1B* (sequencing, MLPA)       *GCK* (sequencing, MLPA)

#### *Porphyria; recessive*

- ALAD* (Doss porphyria) (sequencing)

Please use our separate request form for **postnatal array CGH testing** in patients with mental retardation / multiple congenital anomalies with developmental delay

### Sample and shipping requirements

5-10 ml EDTA blood /  $\geq 10$  ml amniotic fluid / chorionic villi /  $\geq 500$  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).