



www.uk-koeln.de/humangenetik Phone: +49- (0)221- 478 86811

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

Request for molecular genetic analysis

See page 2 for available tests

Billing: Test will be paid by patient referring facility

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

PATIENT INFORMATION (use sticker, if available)
Last name _____
First name(s) _____
Date of birth _____
Address _____ _____
female <input type="checkbox"/> male <input type="checkbox"/>
Ethnic background _____ (may be important in recessive conditions)

Reason for testing:

Please provide pedigree / clinical findings / details on pregnancy (week), previous genetic tests performed in the family, 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.) **Remaining sample material** may be helpful for future diagnostic testing in the proband / in other family members but also for general research/quality management purposes. I consent that remaining material

a) will be stored and may be used for further testing to establish a diagnosis with respect to the above mentioned disorder **yes** **no**

b) will be stored and may be used without disclosing personal data (i.e. in pseudonymized form) for scientific/quality management purposes. **yes** **no**

3.) **Genetic test results** must be destroyed after 10 years according to the German Genetic Diagnostic Act, unless their destruction violates the proband’s interests. I herewith consent that the genetic report will be retained in accordance with privacy protection provisions until I submit a written request for its destruction. **yes** **no**

4.) **Results** of the above mentioned genetic diagnostic testing may be disclosed to:
the following attending physician(s) **yes** **no**

other physicians for counseling family members on the disorder **yes** **no**

*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.*

_____ (Signature referring physician) (Place/Date)

_____ (Name & address physician , phone/fax!)

_____ (Signature patient/legal guardian) (Place/Date)

_____ (Name & address patient/legal guardian , phone)

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

Molecular genetic request form

Neuromuscular Disorders

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

- SMN1* deletion test (MLPA)
- SMN1* carrier test (MLPA)
- SMN1* point mutation analysis (sequencing) (prior enquiry necessary)
- SMN2* (MLPA)

X-linked SMA; X-dominant

- UBE1* (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, hot spot)
- MYBPC1* (sequencing)

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

- RAPSN*
- CHRNA3*
- Other (upon enquiry)

Congenital myopathy (fiber-type disproportion); dominant

- ACTA1* (sequencing)
- SEPN1* (sequencing)
- Other (upon enquiry)

Pontocerebellar hypoplasia (PCH 2 and 4); recessive

- TSEN54*
- TSEN2*
- TSEN34*
- Other (upon enquiry)

Contact : brunhilde.wirth@uk-koeln.de, tel.: -86464; anne.baasner@uk-koeln.de, tel.: -86826; jutta.becker@uk-koeln.de; tel.: -86835; raoul.heller@uk-koeln.de, tel.: -86832

Hearing disorders

Autosomal recessive / digenic hearing loss

- Connexin 26* (sequencing)
- Connexin 30* (PCR of junction fragment)

Pendred syndrome / DFNB4; recessive

- SLC26A4* (sequencing)

Contact : christian.netzer@uk-koeln.de, tel.: -86811; jutta.becker@uk-koeln.de, tel.: -86835; guntram.borck@uk-koeln.de

Craniofacial malformation syndromes

LADD syndrome, ALSG syndrome; dominant

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

Syndromic craniosynostosis; dominant (incl. Apert, Pfeiffer, Crouzon, Saethre-Chotzen, Muenke syndromes)

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

Isolated craniosynostosis

- TWIST* (sequencing)
- TWIST* (MLPA)

Contact: bernd.wollnik@uk-koeln.de, tel.: -86817

Osteogenesis imperfecta

OI type I-IV; dominant

- COL1A1* (sequencing)
- COL1A2* (sequencing)

OI type IIB and VII; recessive

- Testing upon enquiry

Contact: jutta.becker@uk-koeln.de, tel: -86835; christian.netzer@uk-koeln.de, tel: -86811

Cystic fibrosis

Cystic fibrosis; recessive

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

Contact: christian.netzer@uk-koeln.de, tel.: -86811; jutta.becker@uk-koeln.de, tel.: -86835

Kidney disorders

Primary hyperoxaluria type 1 (PH I); recessive

- AGXT* (sequencing)

Primary hyperoxaluria type 2 (PH II); recessive

- GRHPR* (sequencing)

Nephrotic syndrome; recessive

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

Cystic nephropathies/Urinary tract malformations; dominant

- UMOD* (sequencing)
- HNF1 β* (sequencing)

Contact: anne.baasner@uk-koeln.de, tel.: -86826; bodo.beck@uk-koeln.de, tel.: -4319

Sample and shipping requirements: 5-10 ml EDTA blood/ ≥ 10 ml amniotic fluid / CVS / ≥ 500 ng DNA
Ship samples at room temperature.

1 - 2 ml EDTA blood acceptable for newborns and infants (please contact us). 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 (see page 1). → → Please contact us before submitting samples for prenatal diagnosis/during pregnancy. ← ←