UNIVERSITÄTS KLINIKUM HEIDELBERG

Institut für Humangenetik I Im Neuenheimer Feld 366 I 69120 Heidelberg

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DAkkS Deutsche Akkreditierungsstelle D-ML-13060-03-00

Request Form Molecular Genetics

Enclosed sample of: female ☐ male ☐ Date of sample collection: **Family name:** **First name:** **Date of birth:** **Address:** **Patient ethnicity:**	**Sender´s name and direction:** Hospital / Ward / Outpatients Department / Name of Physician (stamp) Phone no.: Fax no:

Coverage of costs:	☐ Public Health Insurance / Outpatient (with referral letter)	☐ Public Health Insurance / Inpatient	☐ Private Health Insurance / Outpatient	☐ Private Health Insurance / Inpatient	☐ Self-paying patient

Self-paying patients / patients with public health insurance without ´Überweisungsschein´ / private health insurance:
I am aware that all costs will have to be covered by myself and I explicitly declare that I assume full responsibility for all costs arising out of my treatment and care. Date:……Signature:……
I have notified my health-insurer and my medical expenses will be covered. Date:……Signature:……

Patient Information

Indication:

If applicable, pedigree //description of symptoms //details on pregnancy (week of gestation) // if applicable, copies of previous medical reports
Key disease symptoms: 1. 2. 3. 4,
Consanguinity: ☐ yes ☐ no
Status post stem cell transplantation: ☐ yes ☐ no

Additional information:

☐ Please complete if molecular genetic testing has been carried out in your family previously:

Family name or name of index patient: _____

When and where? _____ (in case of external analyses, medical report must be attached)

☐ Samples of other family members: ☐ have already been dispatched: ☐ will follow:

Family name, First name,	Date of birth	Degree of kinship	Affected?
_____	_____	*mother*	_____
_____	_____	*father*	_____
_____	_____	*further child(ren)*	_____

Name of treating physician:.....**Phone no:**.....
(capital letters)

……
Date and signature of treating physician

See next pages for request **Please do not forget to fill in informed consent!**

Please tick appropriate box(es)

☐ **Storage of DNA specimen** (an appropriate declaration of consent is required – without it, sample material will be destroyed after one month)

☐ **Testing for maternal contamination of fetal sample in prenatal diagnosis**

☐ **Exome-wide analysis based on single whole genome sequencing including genome-wide detection of deletions and duplications** # ► blood samples from patient **and** his / her parents are required. Please provide detailed medical reference information including key disease symptoms and / or HPO terms (https://hpo.jax.org/app/) and, if possible, a recent medical report

☐ **Exome-wide analysis based on trio whole genome sequencing including genome-wide detection of deletions and duplications** # ► blood samples from patient **and** his / her parents are required. Please provide detailed medical reference information including key disease symptoms and / or HPO terms (https://hpo.jax.org/app/) and, if possible, a recent medical report.

☐ **Preimplantation genetic diagnosis (PGD) ►Prior notification is required**

- ☐ Identification of family-specific polymorphic markers linked to the disease-related gene
- ☐ Testing of the established family-specific (likely) pathogenic variant detection strategy on single cells (leucocytes)

Neuropediatric and other disorders:

☐ **Angelman syndrome**

- ☐ incl. chromosome analysis

☐ **Azoospermia (AZF)** #

- ☐ incl. chromosome analysis

Cystic fibrosis (CF) and *CFTR* related diseases (incl. obstructive azoospermia) (*CFTR* gene)

Patient ethnicity_____ (important for risk calculation)

- ☐ Screening for 50 most frequent pathogenic *CFTR* variants
- ☐ Complete gene analysis (Sanger sequencing of the coding region and MLPA)

☐ **DMD/BMD Muscular dystrophy type Duchenne or type Becker (*DMD* gene)**

☐ **Fragile X syndrome (*FMR1* gene)**

- ☐ incl. chromosome analysis

Hereditary amyloidosis

- ☐ Transthyretin (*TTR* gene)
- ☐ Apolipoprotein A-I (*APOA1* gene)
- ☐ Fibrinogen alpha (*FGA* gene)

Huntington disease (repeat number in the *HTT* gene)

- ☐ test of symptomatic individual
- ☐ presymptomatic test (genetic counseling is mandatory prior to the testing)

Marfan syndrome and other connective tissue diseases #

(virtual multigene panel based on whole genome sequencing)

- ☐ Marfan syndrome und type 1 fibrillinopathy (genes: *FBN1*, *TGFBR1*, *TGFBR2*)
- ☐ Thoracic aortic dilatations (genes: *ACTA2*, *COL3A1*, *FBN1*, *MYH11*, *MYLK*, *SMAD3*, *TGFB2*, *TGFBR1*, *TGFBR2*)

☐ **Prader-Willi syndrome**

- ☐ incl. chromosome analysis

Pulmonary arterial hypertension (PAH)[1] / Osler-Rendu-Weber syndrome (Hereditary hemorrhagic telangiectasia; HHT)[2] / Pulmonary Veno-Occlusive Disease (PVOD)[3] (MGPS)

- ☐ Core genes: *BMPR2*[1,2], *ACVRL1(ALK1)*[1,2], *ENG*[1,2], *EIF2AK4*[1,3]
- ☐ Genes of extended panel: *ABCC8*[1], *AQP1*[1], *ATPA13A3*[1], *BMPR1B*[1], *CAV-1*[1], *GDF2 (BMP9)*[1,2], *KCNA5*[1], *KCNK3*[1], *KDR*[1], *KLF2*[1], *SMAD4*[1,2-], *SMAD5*[1,2], *SMAD9*[1], *SOX17*[1], *TBX4*[1]

☐ **Spinal muscular atrophy (SMA) (copy numbers exon 7 of the *SMN1* and *SMN2* gene)**

Uniparental disomy / Microsatellite analysis ► blood samples of patient **and** his / her parents are required

- ☐ UPD chromosome 7
- ☐ UPD chromosome 14
- ☐ UPD chromosome 15
- ☐ UPD chromosome X

Pharmacogenetics:

☐ Testing for the following clinically relevant variants in *DPYD* gene due to (planned) 5-fluorouracil-based therapy:
c.1129-5923C>G (Haplotyp B3), c.1679T>G (*DPYD**13), c.1905+1G>A (*DPYD**2A), c.557A>G, c.868A>G, c.2279C>T, c.2846A>T

#: non-accredited analysis

Metabolic disorders:

- ☐ Congenital adrenal hyperplasia (21-Hydroxylase deficiency; CAH) (*CYP21A2* gene)
- ☐ Glutaric aciduria type I (*GCDH* gene)
- ☐ MCAD deficiency (*ACADM* gene)
- ☐ Ornithine transcarbamylase deficiency (*OTC* gene)
- ☐ Phenylketonuria/ Hyperphenylalaninemia (*PAH* gene)
- ☐ Hyperphenylalaninemia (*DNAJC12* gene) #
- ☐ Smith-Lemli-Opitz syndrome (*DHCR7* gene)
- ☐ Fabry disease (*GLA* gene) #

Hereditary tumor diseases:

In case of predictive testing, please enclose a copy of original medical report.

Autosomal recessive adenomatous polyposis (MAP)

- ☐ *MUTYH* gene (sequencing and MLPA)
- ☐ test for familial (likely) pathogenic variants

Familial adenomatous polyposis (FAP)

- ☐ *APC* gene (sequencing and MLPA)
- ☐ test for familial (likely) pathogenic variants

Familial breast- and ovarian cancer (MGPS)

- ☐ *BRCA1, BRCA2, RAD51C, RAD51D, CHEK2, PALB2, ATM, BRIP1, BARD1, CDH1,* and *TP53 gene* (gene panel sequencing), MLPA (*BRCA1* and *BRCA2* gene)
- ☐ *BRCA1, BRCA2, RAD51C, RAD51D, CHEK2, PALB2, ATM, BRIP1, BARD1,* and *CDH1 gene* (gene panel sequencing), MLPA (*BRCA1* and *BRCA2* gene)
- ☐ *BRCA1* and *BRCA2* gene (gene panel sequencing and MLPA)
- ☐ additional genes associated with ovarian/ colorectal cancer: *MLH1*, *MSH2*, *MSH6* (gene panel sequencing and MLPA), *EPCAM* (MLPA)
- ☐ test for familial (likely) pathogenic variants

Hereditary nonpolyposis colorectal cancer (HNPCC) *MLH1, MSH2, MSH6* gene (MGPS) #

MSI- / Immunohistochemical analysis should be completed prior to molecular testing (please enclose copy of original report).

- ☐ *MLH1* gene (sequencing and MLPA)
- ☐ *MSH2* gene (sequencing and MLPA)
- ☐ *MSH6* gene (sequencing and MLPA)
- ☐ test for familial (likely) pathogenic variant

Multiple endocrine neoplasia type1 (MEN1) #

- ☐ *MEN1 gene* (sequencing and MLPA)
- ☐ test for familial (likely) pathogenic variant

Multiple endocrine neoplasia Typ2 (MEN2) #

- ☐ *RET* gene (sequencing of exons 5, 7, 8, 10,11,13, 14, 15, 16)
- ☐ test for familial (likely) pathogenic variant

Tuberous sclerosis (TSC) #

- ☐ *TSC1* and *TSC2* gene (sequencing and MLPA)
- ☐ test for familial (likely) pathogenic variant

#: non-accredited analysis

Type of specimen: 3-7 ml ml EDTA blood (children 1-3 ml) or DNA
(in case of karyotyping 5-7 ml NH_4 Heparin blood)

Please make sure to properly label the sample tube with the patient´s name and date of birth and send specimen at room temperature to:

Laboratory for Molecular Genetic Diagnostics
Institute of Human Genetics
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69120 Heidelberg
Germany

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Informed consent to genetic testing according to the Genetic Diagnostics Act (GenDG)

Patient / Person being tested

.....
Family name, first name Date of birth

.....
Address

I have been informed about the significance and consequences of the planned genetic analyses and I have had sufficient time for questions and reflection. I have received a patient information sheet (see QR code). I have no further questions. I am aware that I can revoke my consent at any time.

With my signature, I consent / confirm consent on behalf of my relatives / the person for whom I have custody, to the collection of the necessary blood / tissue samples and agree that the findings may be stored in the UKHD patient data system as an aid to diagnosis:

.....

In the course of the planned examination(s), the analysis /analyses may reveal genetic alterations that are not directly related to the indication of the analysis. Such incidental findings may be medically relevant – or possibly become relevant later in life – however, receiving a genetic test result may also cause distress, be burdensome and / or have implications for your life and future. **As a person capable of giving consent, I would like to be informed about incidental findings concerning myself / the person in my custody.**	❒ yes ❒ no
Specificities when performing genetic testing on children: **I would like to be informed about incidental findings that are of potential clinical significance for my child during childhood / adolescence.** Incidental findings will always be reported - at the discretion of the competent physician - if withholding or failure to act upon them would result in harm to the child. **I would also like to be informed about incidental findings that will only be of clinical significance for my child during adulthood (adult-onset conditions)**	❒ yes ❒ no ❒ yes ❒ no
The GenDG stipulates that sample material should be destroyed when no longer required for the testing for which it was requested. **I consent to the storage of the sample material and its use for results verification, future genetic analyses of myself and within the context of my family and for quality assurance.**	❒ yes ❒ no
Surplus material is an important source for quality assurance and for scientific purposes; it is kept encoded, which makes it impossible for unauthorized individuals to attribute the sample to you / your relatives / the person in your custody. **I consent to the use of remaining sample material to aid medical teaching and research.**	❒ yes ❒ no
I allow that the medical and genetic data that has been collected from me / my relatives / the person for whom I have custody, may be used for scientific purposes in a (partially) coded form and under anonymized conditions be published in scientific journals.	❒ yes ❒ no
The GenDG stipulates that results of genetic analyses ought to be destroyed after 10 years. However, this data could become important for you / your child / the person in your custody and other family members in the future. **I agree to the storage of genetic data and analysis results beyond the legally defined period.**	❒ yes ❒ no

....
Place, date **Signature of patient / person to be examined / legal representative**

.....
Name of Treating Physician Signature of Treating Physician

Nur für Privatpatienten

Kostenübernahmeerklärung

Mir ist bewusst, dass ich für alle anfallenden Kosten selbst aufkommen muss und ich erkläre mich ausdrücklich bereit, diese zu begleichen. Sollte ich bei Einreichung der Rechnung bei meiner privaten Krankenversicherung/Krankenkasse einen Teil der Kosten nicht erstattet bekommen, erkläre ich mich ausdrücklich bereit, die Restkosten im vollen Umfang selbst zu bezahlen.

Information und Einverständnis zur gemeinsamen Abrechnung nach GOÄ durch die Firma unimed

Ich bin informiert und einverstanden, dass die Liquidation privat- bzw. wahlärztlicher Leistungen und Laborleistungen des Institutes für Humangenetik der Universitätsklinikums Heidelberg durch das externe Abrechnungsunternehmen unimed Abrechnungsservice für Kliniken und Chefärzte GmbH, Michael-Uwer-Straße 17 - 19, 66687 Wadern, kurz unimed erfolgt.

Einverständnis zur Datenweitergabe an die Firma unimed

Weiterhin gebe ich die freiwillige und jederzeit widerrufliche Einwilligung, dass das Universitätsklinikum Heidelberg bzw. die liquidationsberechtigten Ärzte die erforderlichen patientenbezogenen persönlichen Behandlungsdaten an dieses Unternehmen ausschließlich zur Rechnungsstellung weitergeben dürfen. Insofern entbinde ich das Universitätsklinikum Heidelberg bzw. die zur Liquidation berechtigten Ärzten ausdrücklich von ihrer ärztlichen Schweigepflicht. Die Mitarbeiter von unimed sind zur Vertraulichkeit im Umgang mit Ihren Daten verpflichtet und unterliegen (auch nach Beendigung ihres Beschäftigungsverhältnisses) der Verschwiegenheitsflicht nach §203 StGB. Ansonsten wird auf die Informationen gemäß Artikel 13 und 14 der Europäischen Datenschutzgrundverordnung (DS-GVO) hingewiesen.

Name der Patienten in Druckbuchstaben	Geburtsdatum
Name des Hauptversicherungsnehmers	Geburtsdatum

Ort	Datum	Unterschrift des/der Patienten/Patientin bzw. Unterschrift des/der Hauptversicherungsnehmers