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Akkreditiert nach DIN EN ISO 15189

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Labor für Cytogenetische Diagnostik
Prof. Dr. rer. nat. J. W. G. Janssen: Tel.: 06221-56-39571

Labor für Molekular-Cytogenetische Diagnostik
Prof. Dr. sc. hum. A. Jauch: Tel.: 06221-56-5407

Request Form Molecular Genetics

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

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

For self-paying patients / Patients insured in a Public Health Insurance scheme without referral letter:
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 associated with all necessary servicing. Date: Signature:

Patient Information

Indication: If applicable, pedigree // Description of symptoms // Details on pregnancy (week of gestation) // If applicable, copies of previous reports

Additional information:

☐ if molecular genetic testing has already been carried out in the family:

Family name or name of the index patient: _____

When and where? _____ (in case of external analysis report must be attached)

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

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

Name of responsible physician:.....**Phone no:**.....
(block letters)

……

Date and signature of responsible physician

See next pages for request **Please fill in informed consent enclosed!**

Informed consent for genetic testing (according to GenDG)

Dear patient,
In order to evaluate or clarify the diagnosis specified below, it is considered to perform molecular genetic testing on you / your child. According to the German Genetic Diagnosis Act (GenDG) prior to any genetic analysis detailed medical information is required. Generally, written informed consent has to be obtained from every patient.
For your information please read the following and mark the appropriate answers.

Name: .....

Date of birth: .....

Address: .....

.....

Molecular genetic testing examines the genetic material (DNA) with respect to genetic alterations which could be causative for the disease / disorder that has occurred or has been suspected in you or any of your family members. In case of a suspected diagnosis for a particular disease, the respective gene / genes will be examined. If the clinical diagnosis cannot be restricted to a particular disorder, many different genetic variants will be analysed and detected simultaneously (e.g. by microarray analysis), so as to obtain a genome wide overview.

In case a disease-causing genetic variant (e.g. mutation) is detected, the diagnosis can generally be considered very reliable. If no genetic variation (mutation) can be identified as the cause of your disease, there is still a possibility of a causative mutation in the examined gene / genes or in one or several other genes. Hence, a genetic disease or predisposition to a disorder generally cannot be fully excluded. In such case, we will try to estimate the probability for you or your relatives to be at risk of developing a disease i.e. of being predisposed to a particular genetic condition. Occasionally, the clinical relevance of detected genetic variations remains uncertain. In this case, it will be mentioned in the diagnosis and be discussed with you. Unfortunately, it is not possible to provide detailed information about all genetic variations that may be mainly or to some extent causative for your disease as well as to exclude every single risk for you or your relatives (especially for your children) by means of a genetic analysis. In case that several family members wish to undergo genetic testing it is absolutely essential for us to know the true biological relationship between the individual family members for correct interpretation of the diagnosis. Should the interpretation of a genetic analysis lead to doubts regarding the accuracy of the provided information about the kinship, this will not be disclosed to you unless it is relevant in counselling for the reason for which the sample was submitted.

Please note that there is an inherent minimal risk of confusion when handling samples and performing a laboratory analysis, which can never be completely excluded. However, all precautions are taken to avoid this or other mistakes.

Please read the following carefully and confirm your consent by signing below:

My physician has informed me about the significance and consequences of the genetic examination mentioned below and has given me adequate time to think my decision over. I understand that I can withdraw my consent at any time.

With my signature I consent to the genetic tests for me / my child and the sampling of blood or tissue necessary to clarify the specified diagnosis:

.....

To pass on the analysis report to additional practitioners requires your consent I consent to my results being sent to the following practitioner(s): Name:……	❒ yes ❒ no
I am interested in additional findings, which may arise in the context of array diagnostics or multi gene panel sequencing (MGPS).	❒ yes ❒ no
According to the German Genetic Diagnosis Act (GenDG) any sample material must be destroyed after completion of the genetic test. Only with your expressed consent, it may be stored longer. Surplus sample material may be required to verify some results (follow-up testing), as well as for necessary quality controls. I consent to my sample being stored for follow-up testing, for future new diagnostic possibilities with regard to the above mentioned medical question / problem, and for quality controls.	❒ yes ❒ no
Surplus material may be an important source for further research and development in the field of medical genetic diagnostics. However, before such use, samples will be anonymised and coded in a way which makes it impossible for any other party to track the sample back to an individual. I consent to my coded sample being stored and used in future research.	❒ yes ❒ no
According to the German Genetic Diagnosis Act (GenDG) any family data on you / your child and any genetic data must be destroyed after 10 years. However, these results may later on become important for your children or grandchildren. I consent to my family data / test results / data of my child being stored beyond this period foreseen by law to enable future testing or counseling of my family members.	❒ yes ❒ no

....

Date, Place **Signature of the patient / of the legal representative**

Please tick appropriate box(es)

☐ **Storage of DNA specimen**

☐ **Array diagnostics** (Genome wide sceening for deletions und duplications)

In case the parents or further children are to be included in the analysis, please make sure to attach a separate informed consent and a letter of referral for both parents as well as for every child tested.

☐ **MLPA to confirm Array results ***

☐ **Preimplantation genetic diagnosis (PGD)** **announcement required**

☐ Identification of family-specific polymorphic markers linked to the disease-related gene

☐ Testing of the established family-specific mutation detection strategy on single cells (leucocytes)

Neuropediatric und other disorders:

☐ Angelman syndrome

☐ incl. chromosome analysis

☐ Azoospermia (AZF) *

☐ incl. chromosome analysis

☐ Huntington´s disease (number of repeats in the *HTT* gene)

☐ test of symptomatic individual

☐ presymptomatic test (genetic counseling mandatory prior to testing)

☐ Cystic fibrosis (CF) (*CFTR* gene)
Ethnic origin of the patient_____ (important for risk calculation)

☐ CBAVD (CAVD) diagnostics (atypical CF, male infertility) (*CFTR* gene)
Ethnic origin of the patient_____ (important for risk calculation)

☐ incl. chromosome analysis

☐ Muscular dystrophy type Duchenne (DMD) or type Becker (BMD)

☐ Fragile X syndrome (*FMR1* gene)

☐ incl. chromosome analysis

☐ Hereditary amyloidosis

☐ Transthyretin (*TTR* gene)

☐ Apolipoprotein A-I (*APOA1* gene)

☐ Fibrinogen alpha (*FGA* gene)

☐ Leri-Weill syndrome / short stature (*SHOX* gene) *

☐ Nephrotic syndrome

☐ Nephrin (*NPHS1* gene)

☐ Podocin (*NPHS2* gene)

☐ Wilms tumor (*WT1* gene, exons 8 and 9)

☐ Pelizaeus-Merzbacher disease (*PLP1* gene) *

☐ Prader-Willi / Angelman disease

☐ incl. chromosome analysis

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

☐ Core genes: *BMPR2*[1,2], *ACVRL1(ALK1)*[1,2], *EIF2AK4*[1,3], *ENG* gene[1,2] incl. MLPA (*BMPR2*, *ACVRL1(ALK1)*, *ENG* gene)

☐ Additional genes: *BMPR1B*[1], *CAV1*[1], *GDF2 (BMP9)*[1,2], *KCNA5*[1], *KCNK3*[1], *KLF2*[1], *SMAD4*[1,2], *SMAD9*[1], *TBX4* gene[1]

☐ Rett-syndrome (*MECP2* gene)

☐ Uniparental disomy / Microsatellite analysis blood samples of the patient and his parents are required

☐ UPD chromosome 7

☐ UPD chromosome 14

☐ UPD chromosome 15

☐ UPD chromosome X

☐ X-inactivation assay (Humara (*AR*),*PCSK1N, ZDHHC15, SLITRK4*, depending on informativity of the markers) *

Neurotransmitter disorders and Pterin metabolism defects:

☐ Tyrosin hydroxylase deficiency *(TH* gene*)* *

☐ Aromatic L-amino acid decarboxylase deficiency *(DDC* gene*)* *

*) asterisk: non accredited analysis

☐ 6 Pyruvoyl-tetrahydropterin synthetase deficiency (BH_4-deficiency) *(PTS* gene*)* *
☐ Dihydropteridin reductase deficiency (BH_4-deficiency) *(QDPR* gene*)* *
☐ GTP cyclohydrolase I deficiency (BH_4-deficiency) *(GCH1* gene*)* *
☐ Sepiapterin reductase deficiency (BH_4-deficiency) *(SPR* gene*)* *

Metabolic disorders:

☐ Congenital adrenal hyperplasia (21-Hydroxylase deficiency; CAH) (*CYP21A2* gene)
☐ Glutaric aciduria type I (*GCDH* gene)
☐ Homocystinuria (*CBS* gene) *
☐ LCHAD deficiency, mutation E510Q (*HADHA* gene) *
☐ MCAD deficiency (*ACADM* gene)
☐ Ornithine transcarbamylase deficiency (*OTC* gene)
☐ Phenylketonuria/Hyperphenylalaninemia (*PAH* gene)
☐ Hyperphenylalaninemia (*DNAJC12* gene) *
☐ Smith-Lemli-Opitz syndrome (*DHCR7* gene)
☐ Methylmalonic aciduria (*MUT* gene) *
☐ Methylglutaconic aciduria type 1 (*AUH* gene) *
☐ Fabry disease (*GLA* gene) *

Hereditary cancer syndromes:

Mutational screening:

Predictive testing/ mutational testing: (please enclose copy of original report)

Autosomal recessive adenomatous polyposis (MAP)
- ☐ complete mutational analysis in the *MUTYH* gene

Familial adenomatous polyposis (FAP)
- ☐ complete mutational analysis in the *APC* gene
- ☐ test for familial mutation
- ☐ Deletion-/ Duplications analysis (MLPA)

Familial breast- und ovarian cancer (MGPS)
- ☐ complete mutational analysis in the *BRCA1, BRCA2, RAD51C, RAD51D, CHEK2, PALB2, ATM, BRIP1, CDH1, and TP53 gene,* incl. MLPA (*BRCA1* and *BRCA2* gene)
- ☐ complete mutational analysis in the *BRCA1, BRCA2, RAD51C, RAD51D, CHEK2, PALB2, ATM, BRIP1,* and *CDH1 gene,* incl. MLPA (*BRCA1* and *BRCA2* gene)
- ☐ complete mutational analysis in the *BRCA1* and *BRCA2* gene, incl. MLPA (*BRCA1* and *BRCA2* gene)
- ☐ test for familial mutation

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

MSI- / Immunohistochemical analysis should be completed prior to molecular testing
(If possible please attach analysis report)
- ☐ complete mutational analysis in the *MLH1* gene incl. MLPA
- ☐ complete mutational analysis in the *MSH2* gene incl. MLPA
- ☐ complete mutational analysis in the *MSH6* gene incl. MLPA
- ☐ test for familial mutation

Multiple endocrine neoplasia type1 (*MEN1*) *
- ☐ complete mutational analysis in the *MEN1* (*Menin*) *gene incl. MLPA*
- ☐ test for familial mutation

Multiple endocrine neoplasia Typ2 (*MEN2*) *
- ☐ complete mutational analysis in the *RET* gene
- ☐ test for familial mutation

Tuberous sclerosis (*TSC*)
- ☐ complete mutational analysis in the *TSC1* and *TSC2* gene incl. MLPA
- ☐ test for familial mutation

*) asterisk: non accredited analysis

Type of specimen: **5-10 ml EDTA blood (children 3-5 ml) or DNA**
(in case of karyotyping 3-5 ml NH_4 Heparin blood)

Please mark type of specimen with name and date of birth of the patient.

Please send specimen at room temperature to:
Laboratory for Molecular Genetic Diagnostics
Institut für Humangenetik,
Im Neuenheimer Feld 366
69120 Heidelberg
Germany

Further diagnostic requests:
See request forms **„Cytogenetik und Fluoreszenz in situ Hybridisierung (FISH) Diagnostik“ or „Leukämien und Lymphoproliferative Erkrankungen”**