| PATIENT DATA | CLINIC/DEPARTMENT/PRACTICE |
|---|---|
| | |

**Human Genetics – Diagnostics Laboratory for Cytogenetics and Molecular Genetics**
Medical director:
Prof. Dr. med. Dr. Judith Fischer
Please send samples to:
Institut für Humangenetik
Universitätsklinikum Freiburg
Breisacher Straße 33 | D-79106 Freiburg i. Br.
Tel.: +49 (0)761-270-70570

# REQUEST FOR MOLECULAR GENETIC TESTING

## PATIENT DATA

LAST NAME, first name(s): _____

Date of birth: _____ Sex: ☐ male ☐ female Pregnancy: ☐ no ☐ yes

Street: _____ ZIP Code, City: _____

Country: _____

## PAYMENT INFORMATION

Please add in any case a confirmation for cost absorption and indicate the billing address.

For a cost estimate please contact molekulargenetik.humangenetik@uniklinik-freiburg.de .

☐ private health insurance ☐ inpatient ☐ self-pay patient

## SAMPLE TYPE

☐ EDTA-blood ☐ DNA, isolated from (tissue)_____ Amount/Conc.: _____

☐ Tissue, fresh/fixed ☐ other: _____ Date of extraction: _____

## INFORMATION ABOUT THE PATIENT

| Medical indication / history (please add medical reports) | Pedigree: |
|---|---|
| | |

- Is the family consanguineous? ☐ yes ☐ no
- Has there been molecular genetic testing in the patient or the patient's family before? ☐ yes ☐ no
- If yes, what has been tested in whom and what was the result? (please add report, if possible) _____

  _____
- Ethnic origin: _____
- The patient is supposed to be tested for a familial mutation. ☐ yes ☐ no

Mutation: _____ Gene: _____ (please add medical report)

_____ _____

Date Stamp and Signature Requesting Physician

| PATIENT DATA | CLINIC/DEPARTMENT/PRACTICE |
|---|---|
| | |

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## INFORMED CONSENT

I have received a thorough explanation from my physician with regard to the disease, its possible genetic origin, and the significance and limitations of the planned genetic test(s). All my questions have been answered and I have had the necessary time to consider giving my consent. I hereby consent to genetic testing to determine the genetic cause of the below mentioned clinical condition.

Clinical Diagnosis/Symptoms: _____

| | | |
|---|---|---|
| I consent to the storage and usage of my personal and medical data as well as the remaining test material beyond the legally defined period for verification of results and quality assurance. | ☐ yes | ☐ no |
| I consent to the usage of my personal and medical data for research purposes regarding the above mentioned clinical diagnosis/symptoms in a pseudonymized form. | ☐ yes | ☐ no |
| I would like to be informed about relevant test results. | ☐ yes | ☐ no |
| Occasionally, additional test results are incidentally obtained which may or may not be related to the above mentioned clinical diagnosis/symptoms. I would like to be informed about these kinds of test results. | ☐ yes | ☐ no |
| I consent to the storage of my personal and medical data beyond the legally defined period. For this, the data is archived and transmitted to a professional external service provider for a short period of time. | ☐ yes | ☐ no |
| I consent to the usage of my personal and medical data for the purpose of counselling and testing family members regarding the above mentioned clinical condition. | ☐ yes | ☐ no |

I was informed about being able to revoke my consent at any time without providing a reason.

_____ Date

_____ Signature Patient
(Legal Representative for minor child)

| **Requesting Physician** | | **Stamp** |
|---|---|---|
| LAST NAME, First Name | | |
| Street | | |
| ZIP Code, City | | |
| Country | | |
| Phone | | |
| E-Mail | | |
| | | |
| Date | Signature | |

## EYE DISEASES

### CONGENITAL STATIONARY RETINAL DYSTROPHY
- ☐ *GNAT1, RHO, NYX, CACNA1F, TRPM1, LRIT3* | 6 genes

### ACHROMATOPSY
- ☐ *CNGB3, CNGA3, GNAT2, PDE6C, PDE6H* | 5 genes

### CHORIODEREMIA
- ☐ *CHM, RPE65, OAT* | 3 genes

### CORNEAL DYSTROPHY
- ☐ *DCN, CHST6, COL8A2, GSN, KRT12, KRT3, PIKFYVE, SLC4A11, TACSTD2, TGFB1, UBIAD1, ZEB1* | 12 genes

### LEBER'S CONGENITAL AMAUROSIS
- ☐ *GUCY2D, RPE65, AIPL1, CRB1, RPGRIP1, LCA5, NMNAT1, CEP290, IMPDH1* | 9 genes

### LEBER'S HEREDITARY OPTIC NEUROPATHY
- ☐ *MT-ND4, MT-ND1, MT-ND6, MT-ND2, MT-ND4L* | 5 genes

### CHOROID MELANOMA
- ☐ *GNA11, GNAQ;* exon 4 and exon 5 each
- ☐ Chromosome 3 status (microsatellites)

### STARGARDT DISEASE AND MACULAR DYSTROPHIES
- ☐ *ABCA4, BEST1, CNGB3, CRB1, ELOVL4, IMPG1, PROM1, PRPH2, RPGR* | 9 genes

### OPTIC ATROPHY
- ☐ *OPA1, OPA3, MFN2* | 3 genes

### RETINITIS PIGMENTOSA
- ☐ autosomal dominant and X-linked | 10 genes
  *RHO, PRPF31, PRPH2, RP1, IMPDH1, PRPF8 KLHL7, NR2E3, RPGR, RP2*

- ☐ autosomal recessive und X-linked | 13 genes
  *USH2A, ABCA4, RPGR, RP2, PDE6A, PDE6B, RPE65, CNGA1, EYS, CRB1, CERKL, PROM1, PDE6C*

### CONE AND CONE-ROD DYSTROPHY
- ☐ *ABCA4, GUCY2D, RPGR, CRX, PRPH2, RPGRIP1, PROM1, CNGB3, RDH5, ADAM9, AIPL1, CNGA3, PDE6C, CERKL, KCNV2* | 15 genes

### RETINITIS PUNCTATA
- ☐ *RDH5, PRPH2, RHO* | 3 genes

## VASCULAR AND CONNECTIVE TISSUE DISORDERS

### MARFAN SYNDROME
- ☐ *FBN1, TGFBR1, TGFBR2* | 3 genes

### MARFAN-LIKE
- ☐ *ADAMTSL4*, *MED12*, *SKI*, *UPF3B*, *ZDHHC9* | 5 genes

### LOEYS-DIETZ SYNDROME
- ☐ *TGFBR1*, *TGFBR2*, *SMAD3*, *TGFB2* | 4 genes

### BEALS SYNDROME
- ☐ *FBN2*

### THORACIC AORTIC ANEURYSM AND AORTIC DISSECTION
- ☐ *ACTA2, MYH11, MYLK, COL3A1, FBN1, SMAD3, TGFB2, TGFBR1, TGFBR2, MAT2A, MFAP5, PRKG1, FLNA, TGFB3, EFEMP2, FBLN5, SCL2A10* | 17 genes

### EHLERS-DANLOS SYNDROME, CLASSIC TYPE (TYPE I and II)
- ☐ *COL5A1, COL5A2* | 2 genes

### EHLERS-DANLOS SYNDROME, HYPERMOBILE TYPE (TYPE III)
- ☐ *TNXB*

### EHLERS-DANLOS SYNDROME, VASCULAR TYPE (TYPE IV)
- ☐ *COL3A1*

### EHLERS-DANLOS SYNDROME, KYPHOSKOLIOSIS TYPE (TYP VII)
- ☐ *COL1A1, COL1A2* | 2 genes

### EHLERS-DANLOS SYNDROME WITH PERIVENTRICULAR HETEROTOPIA
- ☐ *FLNA*

### EHLERS-DANLOS SYNDROME, RECESSIVE SUBTYPES
- ☐ *ADAMTS2, B3GALT6, B4GALT7, CHST14, COL1A2, DSE, FKBP14, PLOD1, SLC39A13* | 9 genes

# SKIN DISEASES

## 1. ICHTHYOSES

### NON SYNDROMAL ICHTHYOSES

COMMON ICHTHYOSES
- ☐ *FLG, STS* | 2 genes

KERATINOPATHIC ICHTHYOSES
- ☐ *KRT1, KRT10, KRT2* | 3 genes

AUTOSOMAL RECESSIVE CONGENITAL ICHTHYOSES (ARCI)
- ☐ *TGM1, NIPAL4, ALOX12B, CYP4F22, ALOXE3, PNPLA1, CERS3, ABCA12, SDR9C7, SULT2B1, CASP14* | 11 genes

### SYNDROMAL ICHTHYOSES (AUTOSOMAL RECESSIVE)

ENTIRE SUBPANEL
- ☐ *ABHD5 (CGI58), ALDH3A2, AP1S1, MPLKIP, CLDN1, ERCC2, ERCC3, GJB2, GJB6, GTF2H5, PHYH, POMP, SLC27A4, SNAP29, SPINK5, ST14, SUMF1, VPS33B* | 18 gene

NETHERTON SYNDROME
- ☐ *SPINK5*

ICHTHYOSIS-HYPOTRICHOSIS SYNDROME (IHS)
- ☐ *ST14*

NEONATAL ICHTHYOSIS-HYPOTRICHOSIS-SCLEROSING CHOLANGITIS SYNDROME (IHSC)
- ☐ *CLDN1*

TRICHOTHIODYSTROPHY
- ☐ *GTF2H5, ERCC2, ERCC3, MPLKIP* | 4 genes

KID SYNDROME (KERATITIS-ICHTHYOSIS-DEAFNESS)
- ☐ *GJB2, GJB6* | 2 genes

ICHTHYOSIS-PREMATURITY SYNDROM (IPS)
- ☐ *FATP4 (SLC27A4)*

SJÖGREN-LARSSON SYNDROME
- ☐ *ALDH3A2*

REFSUM SYNDROME (HMSN4)
- ☐ *PHYH*

MEDNIK SYNDROME
- ☐ *AP1S1*

CEDNIK SYNDROME
- ☐ *SNAP29*

ARTHROGRYPOSIS-RENAL DYSFUNCTION-CHOLESTASIS SYNDROME
- ☐ *VPS33B*

MULTIPLE SULFATASE DEFICIENCY
- ☐ *SUMF1*

CHANARIN-DORFMAN SYNDROME
- ☐ *ABHD5 (CGI58)*

KLICK SYNDROME
- ☐ *POMP*

### SYNDROMAL ICHTHYOSES (X-CHROMOSOMAL)

ENTIRE SUBPANEL
- ☐ *STS, MBTPS2, PORCN, NSDHL, EBP* | 5 genes

SYNDROMAL X-CHR. RECZESSIVE ICHTHYOSIS (Deletion analysis including adjacent genes)
- ☐ *STS*

ICHTHYOSIS FOLLICULARIS – ALOPECIA – PHOTOPHOBIA SYNDROME (IFAP)
- ☐ *MBTPS2*

GOLTZ-GORLIN SYNDROME, FOCAL DERMAL HYPOPLASIA
- ☐ *PORCN*

CHILD SYNDROME
- ☐ *NSDHL*

CONRADI-HÜNERMANN-HAPPLE SYNDROME
- ☐ *EBP*

## 2. HYPERKERATOSES UND PEELING-SKIN SYNDROME

MORBUS DARIER
- ☐ *ATP2A2*

SEVERE DERMATITIS – MULTIPLE ALLERGIES – METABOLIC WASTING – SYNDROME (SAM)
- ☐ *DSG1, DSP* | 2 genes

OLMSTED SYNDROME
- ☐ *MBTPS2, TRPV3* | 2 genes

PEELING-SKIN SYNDROME
- ☐ *TGM5, CDSN, SERPINB8, CHST8, CSTA* | 5 genes

## 3. PALMOPLANTAR KERATODERMA

PANEL DIFFUSE PPK
- ☐ *KRT1, KRT9, SLURP1, SERPINB7, AQP5, CARD14, GJB2, TRPV3, MBTPS2, JUP, POMP, CTSC, SERPINB8* | 13 genes

PACHYONYCHIA CONGENITA
- ☐ *KRT6A, KRT6B, KRT16, KRT17* | 4 genes

DYSPLASIA WITH ASSOCIATED PPK
- ☐ *KRT16, KRT17, KRT6A, KRT6B, CTSC, WNT10A, GJB6* | 7 genes

OTHER DISEASES WITH PPK
- ☐ *DSC2, KANK2, MT-TS1, PTEN, GJB4, TAT* | 6 genes

PANEL STRIATE PPK
- ☐ *KRT6C, KRT16, DSG1, DSP, KRT1, RHBDF2, KRT6A, KRT6B, KRT17* | 9 genes

PANEL PUNCTATED PPK
- ☐ *AAGAB, COL14A1, ENPP1* | 3 genes

PPK WITH ERYTHROKERATODERMA (PERIORIFICIAL, SKIN FOLDS)
- ☐ *KDSR* | 1 gene

## 4. EPIDERMOLYSIS BULLOSA

EPIDERMOLYSIS BULLOSA SIMPLEX - SUPRABASAL
- ☐ *TGM5, DSP, JUP, PKP1* | 4 genes

EPIDERMOLYSIS BULLOSA SIMPLEX - BASAL COMMON SUBTYPES
- ☐ *KRT5, KRT14, KLHL24, TGM5* | 4 genes

EPIDERMOLYSIS BULLOSA SIMPLEX - BASAL RARE SUBTYPES
- ☐ *DST, EXPH5, PLEC* | 3 genes

DYSTROPHIC EPIDERMOLYSIS BULLOSA
- ☐ *COL7A1*

JUNCTIONAL EPIDERMOLYSIS BULLOSA
- ☐ *LAMA3, LAMB3, LAMC2, COL17A1* | 4 genes

JUNCTIONAL EPIDERMOLYSIS BULLOSA WITH/WITHOUT PYLORIC ATRESIA
- ☐ *ITGA6, ITGB4* | 2 genes

JUNCTIONAL EPIDERMOLYSIS BULLOSA WITH LUNG AND KIDNEY INVOLVEMENT
- ☐ *ITGA3*

KINDLER SYNDROME
- ☐ *FERMT1*

## 5. OTHER GENODERMATOSES

ERYTHROKERATODERMIA VARIABILIS
- ☐ *GJB3, GJB4, GJA1, CARD14* | 4 genes

PUSTULAR PSORIASIS, PITYRIASIS RUBRA PILARIS
- ☐ *IL36RN, CARD14* | 2 genes

PARKES-WEBER SYNDROME
- ☐ *RASA1*

MONILETHRIX
- ☐ *KRT81, KRT83, KRT86* | 3 genes

AKNE INVERSA
- ☐ *PSEN1, PSENEN, NCSTN* | 3 genes

BROOKE-SPIEGLER SYNDROME
- ☐ *CYLD*

COLE DISEASE
- ☐ *ENPP1*

ROTHMUND-THOMSON SYNDROME
- ☐ *RECQL4*

WHITE SPONGE NEVUS
- ☐ *KRT4, KRT13* | 2 genes

RESTRICTIVE DERMOPATHY
- ☐ *ZMPSTE24, LMNA* | 2 genes

## CARDIAC DISEASES

BRUGADA SYNDROME
- ☐ *SCN5A, CACNA1C, CACNB2, KCNE3, TRPM4, GPD1L, SCN1B, HCN4, KCNJ8, CACNA2D1, KCND3, KCNE5, RANGRF, SCN2B, SCN3B, SLMAP, SCN10A* | 17 genes

CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA (CPVT)
- ☐ *RYR2, CASQ2, CALM1, CALM2, TRDN* | 5 genes

LONG-QT SYNDROME
- ☐ *KCNQ1, KCNH2, SCN5A, CACNA1C, KCNE1, KCNE2, CALM1, CAV3, KCNE3, KCNJ2, KCNJ5, NOS1AP, SCN4B, AKAP9, ANK2, SNTA1* | 16 genes

ATRIAL FIBRILLATION
- ☐ *ABCC9, GJA5, KCNA5, KCNE1, KCNE2, KCNJ2, KCNQ1, NKX2-5, NPPA, SCN2B, SCN3B, SCN4B SCN5A* | 13 Gene

HYPOPLASTIC LEFT HEART SYNDROME
- ☐ *GJA1, NKX2-5* | 2 genes

ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA (ARVD)
- ☐ *PKP2, DSG2, DSP, DSC2, JUP, TGFB3, TMEM43, LMNA, CTNNA3, RYR2* | 10 genes

DILATED CARDIOMYOPATHY (DCM)
- ☐ *LMNA, MYH7, MYBPC3, SCN5A, TNNT2, TNNI3, CAV3, DES, DSG2, PLN, DSP, ABCC9* | 12 genes

HYPERTROPHIC CARDIOMYOPATHY (HCM)
- ☐ *MYBPC3, MYH7, TNNI3, TNNT2, MYL2, PLN, PRKAG2, TPM1* | 8 genes

NON-COMPACTION CARDIOMYOPATHY (NCCM)
- ☐ *MYBPC3, MYH7, TPM1, ACTC1, LDB3, PRDM16, TAZ, TNNT2* | 8 genes

IDIOPATHIC VENTRICULAR FIBRILLATION
- ☐ *DPP6, KCNJ8, SCN5A* | 3 genes

## NEUROLOGICAL DISEASES

PARKINSON DISEASE
- ☐ *PRKN, PINK1, DJ1, DNAJC6, SYNJ1, SNCA, LRRK2, VPS35, EIF4G1, GBA* | 10 genes

DYSTONIA
- ☐ *TH, SPR, TOR1A, TUBB4A, GCH1, THAP1, SGCE, ATP1A3, CIZ1, ANO3, GNAL* | 11 genes

ATAXIA (autosomal recessive)
- ☐ *VLDLR*

HETEROTOPIA
- ☐ *ARFGEF2, FLNA* | 2 genes

CONGENITAL MUSCULAR DYSTROPHY WITH DYSTROGLYCANOPATHY
- ☐ *FKRP, FKTN, LARGE1, POMT1, POMT2* | 5 genes

CORTICAL DYSPLASIA (WITH POLYMICROGYRIA)
- ☐ *TUBB3*

LISSENCEPHALIA
- ☐ *PAFAH1B1, RELN, TUBA1A , NDE1, DCX, ARX, TBR2* | 7 genes

CONGENITAL PRIMARY MICROCEPHALY
- ☐ *ASPM, WDR62, MCPH1, CDK5RAP2, CENPJ CDK6, CEP135, CEP152, PHC1, PNKP, STIL* | 11 genes

POLYMICROGYRIA
- ☐ *GPR56, TUBA8, TUBB2B* | 3 genes

ATYPICAL RETT SYNDROME
- ☐ *FOXG1*

WARBURG MICRO SYNDROME
- ☐ *RAB18, RAB3GAP1, RAB3GAP2* | 3 genes

HUNTINGTON DISEASE
- ☐ *HTT* (Trinucleotide-Repeat-Analysis)

MOEBIUS SYNDROME
- ☐ *HOXB1, TUBB3* | 2 genes

MUSCLE EYE BRAIN DISEASE (MEB)
- ☐ *FKRP, POMGNT1* | 2 Gene

## METABOLIC DISEASES

DIABETES INSIPIDUS

TYROSINEMIA

☐ *AQP2, AVP, AVPR2*

NEUTRAL LIPID STORAGE MYOPATHY
☐ *PNPLA2* (*ATGL*)

MORBUS GAUCHER
☐ *GBA*

☐ *FAH, HPD, TAT*

CYSTIC FIBROSIS
☐ *CFTR*

## TUMOR DISEASES

HEREDITARY BREAST AND OVARIAN CANCER (HBOC)
☐ *BRCA1, BRCA2, CHEK2, PALB2, RAD51C, BARD1*, *BRIP1*, *MRE11A*, *NBN*, *PTEN*, *RAD50*, *RAD51D,TP53, BLM, CDH1, FAM175A*, *MEN1*, *STK11, XRCC2* | 19 genes

HEREDITARY NON-POLYPOSIS COLORECTAL CANCER SYNDROME (HNPCC)
☐ *MLH1, PMS*, *MSH2, MSH6* | 4 genes

FAMILIAL ADENOMATOUS POLYPOSIS (FAP)
☐ *APC, MUTYH* | 2 genes

FAMILIAL PANCREATIC CARCINOMA
☐ *BRCA2, STK11, PALB2, BRCA1, TP53, MEN1 MLH1, PMS2, MSH2, MSH6, ATM* | 11 genes

HEREDITARY DIFFUSE GASTRIC CANCER
☐ *CDH1, MSH6* | 2 genes

OTHER TUMOR PREDISPOSITION SYNDROMES
☐ Li-Fraumeni Syndrome | *TP53*
☐ Birt-Hogg-Dubé Syndrome | *FLCN*
☐ Hamartom-Tumor Syndrome | *PTEN*
☐ Peutz-Jeghers Syndrome | *STK11*
☐ Von-Hippel-Lindau Syndrome | *VHL*

## OTHER DISEASES

AZOOSPERMIA
☐ AZF-Factors

CONGENITAL BILATERAL ABSENCE OF VAS DEFERENS
☐ *CFTR*

CAMPOMELIC DYSPLASIA
☐ *SOX9*

OROFACIODIGITAL SYNDROME
☐ *OFD1*

SENSORINEURAL DEAFNESS (TYPE DFNA and DFNB)
☐ *GJB2* (CX26), *GJB6* (CX30), *GJA1* (CX 43)

NEUROFIBROMATOSIS TYPE I
☐ *NF1*