synlab Medizinisches
Versorgungszentrum
Humane Genetik

Patient data
Last name, first name:

Date of birth:
Referring doctor / clinic:

Ärztliche Leitung:
Dr. med. Dr. rer. nat. Claudia Nevinny-Stickel-Hinzpeter
Fachärztin für Humangenetik

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Order for Molecular Genetic Analyses

Report should be sent to:

e-mail address:

Invoice should be sent to:

e-mail address:

Payment

☐ bank transfer (please see information below)
☐ credit card (please see separate form)
The analysis will be started as soon as the payment is settled.

Gender of the patient: ☐ male ☐ female

Sample material: Collection date: _____

☐ EDTA-blood 2-5 ml ☐ DNA ☐ Other: _____

Clinical data / Diagnosis:

☐ Analysis of a familial mutation

Name and date of birth of index patient: _____

Gene and mutation: _____

If available, please include reports of molecular genetic analysis of affected family members.

Date

Signature and official stamp of the referring physician

Please note: we are continuously expanding our portfolio of analyses. If you do not find a desired analysis on this list please inquire. Analyses not performed in our laboratory will be forwarded to another accredited human genetic laboratory.

Molecular genetic analyses:

Cardiac diseases

- ☐ Arrhythmogenic right ventricular cardiomyopathy/dysplasia (ARVC/D) (*PKP2*)
- ☐ Cardiomyopathy (hypertrophic/dilated)
  - ☐ *MYBPC3* ☐ *MYH7*
  - ☐ *TNNT2* ☐ *TNNI3*
  - ☐ *LMNA*
- ☐ Brugada syndrome, BrS1 (*SCN5A)*
- ☐ Long QT syndrome
  - ☐ LQT1 (*KCNQ1)* ☐ LQT2 (*KCNH2)*
  - ☐ LQT3 (*SCN5A)* ☐ LQT5 (*KCNE1)*
  - ☐ LQT6 (*KCNE2)*
- ☐ Short QT syndrome
  - ☐ SQT1 (*KCNH2)* ☐ SQT2 (*KCNQ1)*

Complex syndromes

- ☐ Aarskog syndrome (FGD1)
- ☐ Angelman syndrome, AS
  - ☐ *SNRPN*-methylation status ☐ *UBE3A*
- ☐ Beckwith-Wiedemann syndrome, BWS
  - ☐ 11p15 methylation status ☐ UPD11
- ☐ Di George syndrome
- ☐ Fragile X syndrome, FraX
  - ☐ FRAXA (*FMR1*) ☐ FRAXE (*FMR2*)
- ☐ HDR syndrome, hypoparathyreodism, deafness and renal insufficiency (*GATA3*)
- ☐ Kallmann syndrome
  - ☐ X-linked type (*KAL1*) ☐ autosomal type (*FGFR1*)
- ☐ LEOPARD syndrome (*PTPN1*)
- ☐ Noonan syndrome
  - ☐ *PTPN11* ☐ *SOS1*
- ☐ Prader-Willi syndrome, PWS
  - ☐ *SNRPN*-methylation status ☐ UPD15
- ☐ Rett syndrome (*MECP2*)
- ☐ Silver-Russell syndrome, SRS
  - ☐ 11p15 methylation status ☐ UPD7
- ☐ Sotos syndrome (*NSD1*)
- ☐ Williams-Beuren syndrome, WBS

Connective tissue disorders

- ☐ Ehlers-Danlos syndrome type I+II (classic)
  - ☐ *COL5A1* ☐ *COL5A2*
- ☐ Ehlers-Danlos syndrome type IV ,vascular (*COL3A1)*
- ☐ Ehlers-Danlos syndrome type VI, Kyphoscoliosis (*PLOD1*)
- ☐ Ehlers-Danlos syndrome type VIIA+B (Arthrochalasia)
  - ☐ *COL1A1* most common mutations
  - ☐ *COL1A2* most common mutations
- ☐ Loeys-Dietz syndrome
  - ☐ *TGFBR1* ☐ *TGFBR2*
- ☐ Marfan syndrome (*FBN1*)
- ☐ Osteogenesis Imperfecta
  - ☐ *COL1A1* ☐ *COL1A2*

Endocrinology

- ☐ Congenital adrenal hyperplasia, CAH, 21-hydroxylase deficiency (*CYP21A2*)
- ☐ Hyperinsulinism
  - ☐ severe neonatal form, autosomal recessive
    - ☐ *ABCC8*/*SUR*1 ☐ *KCNJ11/Kir6.2*
  - ☐ mild form, autosomal dominant
    - ☐ *GCK* ☐ *GLUD1*
- ☐ Hyperproinsulinemia (*INS)*
- ☐ Kallmann syndrome
  - ☐ X-linked type (*KAL1*) ☐ autosomal type (*FGFR1*)
- ☐ obesity (early onset) (*MC4R*)

Eye diseases

- ☐ Leber hereditary optic neuropathy, LHON m.11778G>A, m.3460G>A, m.144484T>A
- ☐ Optic Atrophy 1 (*OPA1*)

Fertility disorders

Habitual miscarriages

- ☐ Factor V (*F5*) Leiden-mutation
- ☐ Mannose binding lecin (MBL)-deficiency (*MBL2*, mutations p.Gly54Asp, p.Gly57Glu, p.Arg52Cys)

Male infertility

- ☐ Azoospermia factor (AZF)
- ☐ Congenital bilateral Aplasia of Vas deferens, CBAVD (*CFTR)*
  - ☐ 36 common mutations ☐ complete gene

Gastrointestinal diseases

- ☐ inflammatory bowel (Crohn) disease (*NOD2,* mutations p.Arg702Trp, p.Gly908Arg, p.Leu1007Profs*2)
- ☐ Celiac disease (DQA1, DQB1)

Hematology

- ☐ α- Thalassemia
- ☐ β- Thalassemia
- ☐ Sickle cell disease
- ☐ Glucose-6-Phosphat-dehydrogenase-deficiency, Favism (*G6PD*)

Hereditary cancer syndromes

- ☐ Familial adenomatous polyposis coli, FAP
  - ☐ *APC* ☐ *MUTYH*
- ☐ Hereditary non-polyposis colon cancer, HNPCC
  - ☐ *MLH1* ☐ *MSH2* ☐ *MSH6*
- ☐ Hereditary Breast/Ovarian Cancer
  - ☐ *BRCA1* ☐ *BRCA2*
  - ☐ *CHEK2* (mutations c.1100delC, c.444+1G>A, c.470T>C)
- ☐ Multiple endocrine neoplasie type I, MENI (*MEN1*)
- ☐ Multiple endocrine neoplasia type II, MENII (*RET*)
- ☐ Neurofibromatosis type I, NF1 (*NF1*)
- ☐ von-Hippel-Lindau syndrome, VHL (*VHL*)

Immune disorders

- ☐ HLA-B27 associated diseases
- ☐ Celiac disease (DQA1, DQB1)

Intersexuality

- ☐ Congenital adrenal hyperplasia, CAH, 21-hydroxylase deficiency (*CYP21A2*)
- ☐ SRY

Kidney diseases

- ☐ Adult dominant polycystic kidney disease, ADPKD
  - ☐ *PKD1* ☐ *PKD2*
- ☐ Alport syndrome
  - ☐ X-chromosomale type (*COL4A5)*
  - ☐ autosomal recessive/dominant type (*COL4A4, COL4A3)*
- ☐ Renal glucosuria (*SLC5A2*)

Liver diseases

- ☐ Crigler-Najjar syndrome type 1 / 2 (*UGT1A1*)
- ☐ Hemochromatosis, hereditary (*HFE*)
  - ☐ mutations C282Y, H63D, S65C
  - ☐ complete gene
- ☐ Hemochromatosis type 2A (*HJV*)
- ☐ Hemochromatosis type 2B (*HAMP*)
- ☐ Hemochromatosis type 3 (*TFR2*)
- ☐ Hemochromatosis type 4 (*SLC40A1*)
- ☐ Hyperbilirubinemia, Gilbert syndrome (*UGT1A1,* promoter TA-repeat)
- ☐ Wilson disease (*ATP7B*)
  - ☐ most common mutation p.His1069Gln
  - ☐ complete gene

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Humane Genetik München, Zweigniederlassung
der synlab MVZ Augsburg GmbH
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Bankleitzahl 600 202 90
IBAN DE 41 6002 0290 0611 0586 91
Swift-Code HYVEDEMM473

Registergericht
Augsburg HRB 24536
UST-ID DE267015644

Lung diseases
- ☐ α1-antitrypsin deficiency, AAT (*SERPINA1*)
  - ☐ Pi*S, Pi*Z ☐ complete gene
- ☐ Cystic fibrosis, CF (*CFTR*)
  - ☐ F508del mutation
  - ☐ 36 common mutations
  - ☐ complete gene

Metabolic diseases
- ☐ Apolipoprotein A1 (*APOA1*)
- ☐ Apolipoprotein B (*APOB*, mutations R3500Q/W, R3531C)
- ☐ Apolipoprotein E (*APOE*, *2*3*4 alleles)
- ☐ Fabry disease, α-Galactosidase-A deficiency (*GLA*)
- ☐ Familial hypercholesterolemia
  - ☐ LDL-receptor (*LDLR*) ☐ *PCSK9*
- ☐ Fructose intolerance, hereditary (*ALDOB*)
  - ☐ most common mutations ☐ complete gene
- ☐ Glucose-6-Phosphat-dehydrogenase-deficiency, Favism (*G6PD*)
- ☐ Hyperinsulinism
  - ☐ severe neonatal form, autosomal recessive
    - ☐ *ABCC8/SUR*1 ☐ *KCNJ11/Kir6.2*
  - ☐ mild form, autosomal dominant
    - ☐ *GCK* ☐ *GLUD1*
- ☐ Hyperproinsulinemia (*INS)*
- ☐ Lactose intolerance (-13910C/T polymorphism of *LPH*)
- ☐ Medium-chain Acyl-CoA-Dehydrogenase deficiency, MCAD (*ACADM*)
  - ☐ most common mutations ☐ complete gene
- ☐ Maturity onset Diabetes of the Young, MODY
  - ☐ MODY 1 (*HNF4A*) ☐ MODY 2 (GCK)
  - ☐ MODY 3 (*HNF1A/TCF1*) ☐ MODY 4 (*IPF1/PDX1*)
  - ☐ MODY 5 (*HNF1B/TCF2*) ☐ MODY 6 (*NEUROD1*)
  - ☐ MODY 7 (*KLF11*) ☐ MODY10 (*INS*)
- ☐ Neonatal Diabetes
  - ☐ *ABCC8 / SUR1* ☐ *KCNJ11 / Kir6.2*
  - ☐ *GCK* ☐ UPD6 ☐ *INS*
- ☐ Osteoporosis
  - ☐ Sp1 Polymorphism (*COL1A1*)
  - ☐ Vitamin D-receptor (*VDR*, B/b polymorphism)
- ☐ Phenylketonuria, PKU (*PAH*)
- ☐ Renal glucosuria (*SLC5A2*)

Mitochondrial disorders
- ☐ CPEO / Kearns-Sayre / Pearson syndrome (large deletion, 1.3-10 kb)
- ☐ MELAS, Diabetes-Deafness syndrome (m.3243A>G)
- ☐ Leber hereditary optic neuropathy, LHON (m.11778G>A, m.3460G>A, m.144484T>A)
- ☐ Leigh-/NARP syndrome (m.8993T>G, m.8993T>C)
- ☐ MERRF syndrome (m.8344A>G)

Neurodegenerative diseases
- ☐ adult-onset dominant leukodystrophy, ADLD (*LMNB1*)
- ☐ CADASIL (*NOTCH3*)
- ☐ Huntington disease (*HTT*)

Neuromuscular diseases / Neuropathies
- ☐ Charcot-Marie-Tooth disease
  - ☐ CMT1A (*PMP22*) ☐ CMT1B (*MPZ*)
  - ☐ CMT2A (*MFN2*) ☐ CMT1X (*GJB1*)
- ☐ Hereditary neuropathy with liability to pressure Palsies, HNPP (*PMP22*)
- ☐ Muscular dystrophy type Duchenne / Becker, DMD/BMD (*DMD*)
- ☐ Myotonic dystrophy
  - ☐ type I, Curschmann-Steinert (*DMPK*)
  - ☐ type II, PROMM (*CNBP*)
- ☐ Spinal muscular atrophy type I / II / III (*SMN1*)
- ☐ Spinal bulbar muscular atrophy, SBMA, Kennedy syndrome (*AR*)

Nutrigenetics
- ☐ Celiac disease (DQA1, DQB1)
- ☐ Fructose intolerance, hereditary (*ALDOB*)
  - ☐ most common mutations ☐ complete gene
- ☐ Lactose intolerance (-13910C/T polymorphism of *LPH*)

Pancreatic diseases
- ☐ Hereditary pancreatitis
  - ☐ *PRSS1* ☐ *SPINK1*
  - ☐ *CFTR*
    - ☐ F508del mutation
    - ☐ 36 common mutations
    - ☐ complete gene

Periodic fevers
- ☐ CINCA/NOMID (*NLRP3*)
- ☐ Familial mediterranean fever, FMF (*MEFV*)
- ☐ FCAS (*NLRP3*)
- ☐ Hyper IgD Syndrome (*MVK*)
- ☐ Muckle-Wells syndrome (*NLRP3*)
- ☐ TNF receptor-associated periodic syndrome (TRAPS, *TNFRSF1A*)

Pharmacogenetics
- ☐ 5-Fluorouracil (5-FU)-related toxicity (*DPD*, exon 14-skipping mutation)
- ☐ Glutathion S-transferase (*GSTM1, GSTT1*)
- ☐ N-Acetyltransferase 2 (*NAT2*)
- ☐ Statin-induced myopathy (*SLCO1B1*), haplotyp SLCO1B1*5
- ☐ Thiopurin-S-methyltransferase (*TPMT*)
- ☐ UDP-Glycosyl-Transferase (*UGT1A1*)

Short stature
- ☐ Disproportionate short stature (*FGFR3*)
  - ☐ Achondroplasia
  - ☐ Hypochondroplasia
  - ☐ Thanatophoric Dysplasia
- ☐ SHOX deficiency (*SHOX*)
- ☐ Silver-Russell syndrome, SRS
  - ☐ 11p15 methylation status ☐ UPD7

Thrombophilia / Atherosclerosis
- ☐ Angiotensin converting enzyme, ACE (*ACE*, del/ins Polymorphism)
- ☐ ß-Fibrinogen (*FGB*, -455G/A)
- ☐ Factor V (*F5*)
  - ☐ Leiden-mutation ☐ H1299R mutation
- ☐ Factor XIII (*F13*, V34L)
- ☐ Glycoprotein Ia (*ITGA2,* c.807C>T)
- ☐ Glycoprotein IIIa (*ITGB3,* L33P)
- ☐ Methylenetetrahydrofolate-reductase (*MTHFR*, c.677C>T, c.1298A>C)
- ☐ Plasminogen activator inhibitor type1, PAI1 (*PAI1*, 4G/5G Polymorphism)
- ☐ Prothrombin, FII (*F2*, g.20210G>A)

Uniparental disomy
- ☐ UPD6 ☐ UPD7 ☐ UPD11
- ☐ UPD14 ☐ UPD15

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Informed consent for Genetic Analyses

After having received information regarding the significance, risks and limitations I hereby agree to the genetic analysis of the following clinical diagnosis or indication

_____
indication

on behalf of myself or the person in my legal custody.

Storage of the samples

According to German law the sample has to be discarded after completion of the final report. In order to allow re-examination, the samples will be stored for an adequate period of time and then disposed (= legal time-span). However, for some samples a longer term storage may be of relevance.

☐ I consider the legal time-span of storage to be sufficient.

☐ I wish my sample to be stored beyond the legal time-span (max. 10 years).

Use of the samples

☐ I allow my anonymised sample to be used for research and quality control purposes.

☐ I allow my sample to be exclusively used for the above mentioned course of analysis.

Storage of the results

According to German law the results of the analysis have to be destroyed after 10 years of storage (= legal time-span). However, the results may be important for human genetic counseling of children or other relatives of the patient after this period of time.

☐ I consider the legal time-span of storage to be sufficient.

☐ I wish the results to be stored beyond the legal time-span.

I have the right to withdraw this consent at any time by contacting the referring physician.

I have the right of refusal to receive the results after the completion of the analysis.

If requested, the results of the analysis can be transmitted per email or fax. I am aware that this mode of transmission may be insecure.

_____
Date, place

_____
Signature of the patient or guardian

_____
Signature and stamp of the referring physician