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 variant

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.

Cardiac diseases
- ☐ Arrhythmogenic right ventricular cardiomyopathy/dysplasia, ARVC/D (*PKP2, DSG2, DSP, DSC2*)
- ☐ Brugada syndrome, BrS1 (*SCN5A)*
- ☐ Jervell-Lange-Nielsen syndrome (*KCNE1, KCNQ1*)
- ☐ Cardiomyopathy, hypertrophic/dilated (*MYBPC3, MYH7, TNNT2, TNNI3, LMNA, MYH6, BAG3, ANKRD1, TMPO, TPM1, MYL3, RBM20, LDB3, TCAP*)
- ☐ Catecholaminergic polymorphic ventricular tachycardia, CPVT (*RYR2*)
- ☐ 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*
- ☐ Costello syndrome (*HRAS*)
- ☐ Di George syndrome
- ☐ Fragile X syndrome, FraX (*FMR1*)
- ☐ Fragile X syndrome, FRAXE (*AFF2*)
- ☐ Hutchinson-Gilford Progeria (*LMNA*)
- ☐ Hypoparathyreodism, Deafness, Renal insufficiency, HDR (*GATA3*)
- ☐ Kallmann syndrome (*KAL1, FGFR1, PROK2, PROKR2*)
- ☐ LEOPARD syndrome (*PTPN11, RAF1, BRAF*)
- ☐ Noonan syndrome
  - ☐ basic analysis type 1 (*PTPN11*)
  - ☐ comprehensive analysis: type 3 (*KRAS*), type 4 (*SOS1*), type 5 (*RAF1*), type 7 (*BRAF*), type 8 (*RIT1*)
- ☐ Osler-Rendu-Weber disease (*ENG, ACVRL1*)
- ☐ Prader-Willi syndrome, PWS
  - ☐ *SNRPN*-methylation status ☐ UPD15*
- ☐ Proteus syndrome (*AKT1*)
- ☐ Rett syndrome (*MECP2*)
- ☐ Silver-Russell syndrome, SRS (11p15 methylation status, UPD7)
- ☐ Sotos syndrome (*NSD1*)
- ☐ Thrombocytopenia, absent radius syndrome, TAR (*RBM8A*)
- ☐ Williams-Beuren syndrome, WBS

Connective tissue disorders
- ☐ Ehlers-Danlos syndrome, classic type (*COL5A1, COL5A2*)
- ☐ Ehlers-Danlos syndrome, vascular type (*COL3A1)*
- ☐ Ehlers-Danlos syndrome, Kyphoscoliosis type(*PLOD1*)
- ☐ Ehlers-Danlos syndrome, Arthrochalasia type (*COL1A1, COL1A2*)
- ☐ Marfan syndrome / Loeys-Dietz syndrome (*FBN1, TGFBR1, TGFBR2*)
- ☐ Osteogenesis Imperfecta (*COL1A1, COL1A2*)
- ☐ Tenascin X-deficiency (*TNXB*)
- ☐ Thoracic aortic aneurysms and aortic dissections, TAAD (*ACTA2, COL3A1, FBN1, MYH11, MYLK, SMAD3, TGFB2, TGFBR1, TGFBR2*)

Endocrinology
- ☐ Congenital adrenal hyperplasia, CAH
  - ☐ 21-hydroxylase deficiency (*CYP21A2*)
  - ☐ Steroid 11-beta-Hydroxylase deficiency (*CYP11B1*)
  - ☐ 17-alpha-Hydroxylase deficiency (*CYP17A1*)
  - ☐ 3-beta-Hydroxysteroid Dehydrogenase 2 deficiency (*HSD3B2*)
  - ☐ P450 Oxidoreductase deficiency (*POR*)

  Hyperinsulinism → please see "Metabolic diseases"
  Hyperproinsulinemia → please see "Metabolic diseases"
  Kallmann syndrome → please see "Complex syndromes"
  MODY → please see "Metabolic diseases"
  Neonatal Diabetes → please see "Metabolic diseases"
- ☐ Obesity, early onset (*MC4R, MC3R, POMC, LEPR, LEP*)

All genes combined to panels may be analysed individually on request.

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

Fertility disorders
Habitual miscarriages
- ☐ Factor V (*F5*) Leiden variant
- ☐ Mannose binding lectin (MBL)-deficiency (*MBL2*)

Male infertility
- ☐ Azoospermia factor (AZF)
- ☐ Congenital bilateral Aplasia of Vas deferens, CBAVD (*CFTR)*
  - ☐ 50 common variants ☐ complete gene

Gastrointestinal diseases
- ☐ Celiac disease (DQA1, DQB1)
- ☐ Inflammatory bowel (Crohn) disease (*NOD2,* variants p.(Arg702Trp), p.(Gly908Arg), p.(Leu1007Profs*2))

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

Hemophilia
- ☐ Hemophilia A (*F8*)
- ☐ Hemophilia B (*F9*)
- ☐ von-Willebrand disease (*VWF*)

Hereditary cancer syndromes
Please see the separate order form for hereditary cancer syndromes on our website.
- ☐ Ataxia teleangiectasia (*ATM*)
- ☐ Familial adenomatous polyposis coli, FAP1 (*APC*), FAP2 (*MUTYH*)
- ☐ Hereditary Breast/Ovarian Cancer, HBOC (*BRCA1, BRCA2, RAD51C, PALB2, CHEK2*)
- ☐ Hereditary diffuse gastric cancer (*CDH1*)
- ☐ Hereditary non-polyposis colon cancer, HNPCC (*MLH1, MSH2, MSH6, PMS2*)
- ☐ Juvenile polyposis syndrome (*BMPR1A, SMAD4*)
- ☐ Li-Fraumeni syndrome (*TP53*)
- ☐ Multiple endocrine neoplasia type I, MEN1 (*MEN1*)
- ☐ Multiple endocrine neoplasia type II, MEN2 (*RET*)
- ☐ Neurofibromatosis, NF
  - ☐ type 1 (*NF1*) ☐ type 2 (*NF2*)
- ☐ Peutz-Jeghers syndrome (*STK11*)
- ☐ PTEN hamartoma tumor syndrome (*PTEN*)
- ☐ Tuberous sclerosis (*TSC2, TSC1*)
- ☐ von-Hippel-Lindau syndrome, VHL (*VHL*)

Immune disorders
- ☐ Behçet syndrome
- ☐ HLA-B27 associated diseases
- ☐ Narcolepsia

Intersexuality
Congenital adrenal hyperplasia, CAH → please see "Endocrinology"
- ☐ SRY

Kidney diseases
- ☐ Autosomal dominant polycystic kidney disease, ADPKD (*PKD1, PKD2)*
- ☐ Autosomal recessive polycystic kidney disease, ARPKD (*PKHD1*)
- ☐ Alport syndrome (*COL4A5, COL4A3, COL4A4*)
- ☐ Renal glucosuria (*SLC5A2*)

Liver diseases
- ☐ Crigler-Najjar syndrome type 1 / 2 (*UGT1A1*)
- ☐ Hemochromatosis
  - ☐ basic analysis type 1 (*HFE*) p.Cys282Tyr, p.His63Asp, p.Ser65Cys
  - ☐ comprehensice analysis: type 1 (*HFE*) complete gene, type 2A (*HFE2*), type 2B (*HAMP*), type 3 (*TFR2*), type 4 (*SLC40A1*)
- ☐ Hyperbilirubinemia, Gilbert syndrome (*UGT1A1,* promoter TA-repeat)
- ☐ Wilson disease (*ATP7B*)
  - ☐ most common variant p.(His1069Gln) ☐ complete gene

SYNLAB Medizinisches Versorgungszentrum Humane Genetik München,
Zweigniederlassung der SYNLAB MVZ Augsburg GmbH – synlab.com
Geschäftsführung: Mathieu Floreani, Doris Schlieszeit, Dr. Christoph Mahnke, Andrea Schikora

HypoVereinsbank Stuttgart
Swift-Code HYVEDEMM473
IBAN DE 41 6002 0290 0611 0586 91

Registergericht
Augsburg HRB 24536
UST-ID DE267015644

Lung diseases
- ☐ α1-antitrypsin deficiency, AAT (*SERPINA1*)
  - ☐ Pi*S, Pi*Z ☐ complete gene
- ☐ Cystic fibrosis, CF (*CFTR*)
  - ☐ p.(Phe508del) variant ☐ 50 common variants
  - ☐ complete gene

Metabolic diseases
- ☐ Apolipoprotein A1 (*APOA1*)
- ☐ Apolipoprotein B (*APOB*, p.(Arg3527Gln/Trp), p.(Arg3558Cys))
- ☐ Apolipoprotein E (*APOE*, alleles E2, E3, E4)
- ☐ Fabry disease, α-Galactosidase-A deficiency (*GLA*)
- ☐ Familial hypercholesterolemia (*LDLR, PCSK9, LDLRAP1*)

Fructose intolerance (*ALDOB*) → please see "Nutrigenetics"
Glucose-6-Phosphat-dehydrogenase-deficiency → see "Hematology"
- ☐ Gaucher disease (*GBA*)
- ☐ Hyperinsulinism (*ABCC8, KCNJ11, GCK, GLUD1, HNF4A*)
- ☐ Hyperproinsulinemia (*INS*)

Lactose intolerance → please see "Nutrigenetics"
- ☐ Lipodystrophy type Dunnigan (*LMNA*)
- ☐ Maturity onset Diabetes of the Young, MODY 1-13 (*HNF4A, GCK, HNF1A, PDX1, HNF1B, NEUROD1, KLF11, CEL, PAX4, INS, BLK, ABCC8, KCNJ11*)
- ☐ Medium-chain Acyl-CoA-Dehydrogenase deficiency, MCAD (*ACADM*)
  - ☐ most common variants ☐ complete gene
- ☐ Neonatal Diabetes, permanent, transient (*ABCC8, KCNJ11, GCK,* UPD6, *INS*)

Obesity, early onset → please see "Endocrinology"
- ☐ Osteoporosis (Sp1 polymorphism *COL1A1*, BsmI polymorphism (*VDR*)
- ☐ Phenylketonuria, PKU/ Hyperphenylalaninemia (*PAH*)
- ☐ Porphyrien: acute intermittent (*HMBS*), variegate (*PPOX*), P. cutanea tarda (*UROD*), erythropoietic protoporphyria (*FECH*), Doss porphyria (*ALAD*), congenital erythropoietic porphyria (*UROS*) X-chromosomale protoporphyri (*ALAS2*), Coproporrphyria (*CPOX*)

Renal glucosuria (*SLC5A2*) → please see "Kidney diseases"

Mitochondrial disorders
- ☐ Chronic progressive external ophthalmoplegia, CPEO, Kearns-Sayre syndrome, KSS, Pearson syndrome

Leber hereditary optic neuropathy → please see "Eye diseases"
- ☐ Leigh- / Neuropathy, ataxia, retinitis pigmentosa (NARP) syndrome (m.8993T>G, m.8993T>C)
- ☐ Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes, MELAS, Diabetes-Deafness syndrome (m.3243A>G)
- ☐ Myoclonic epilepsy associated with ragged-red fibers, MERRF syndrome (m.8344A>G)

Neurodegenerative diseases
Please see the separate order form for neurodegenerative diseases on our website.
- ☐ Autosomal dominant adult-onset demyelinating leukodystrophy, ADLD (*LMNB1*)
- ☐ CADASIL / CARASIL, Cerebral Autosomal dom. /rec. Arteriopathy with subcortical Infarcts and Leukoencephalopathy (*NOTCH3, HTRA1*)
- ☐ Friedreich Ataxia (*FXN*)
- ☐ Huntington disease (*HTT*)
- ☐ Spinocerebellar ataxias (SCA)
  - ☐ SCA1 *(ATXN1)* ☐ SCA2 (*ATXN2*)
  - ☐ SCA3, Machado-Joseph disease, MJD (*ATXN3*)
  - ☐ SCA6 *(CACNA1A)* ☐ SCA7 *(ATXN7)*

All genes combined to panels may be analysed individually on request.

Neuromuscular diseases / Neuropathies
- ☐ Charcot-Marie-Tooth disease
  - ☐ basic analysis CMT1A (*PMP22*)
  - ☐ comprehensive analysis: CMT1B (*MPZ*), CMT1C (*LITAF*), CMT1D (*EGR2*), CMT1X (*GJB1*), CMT2A (*MFN2*), CMT2A1 (*KIF1B*), CMT2B1 (*LMNA*), CMT2D (*GARS*), CMT2E/1F (*NEFL*)
- ☐ Emery-Dreifuss muscular dystrophy, EDMD (*LMNA*)
- ☐ Hereditary neuropathy with liability to pressure Palsies, HNPP (*PMP22*)
- ☐ Limb girdle muscular dystrophy 1B, LGMD1B (*LMNA*)
- ☐ Muscular dystrophy type Duchenne / Becker, DMD/BMD (*DMD*)
- ☐ Myotonic dystrophy
  - ☐ type I, Steinert disease (*DMPK*)
  - ☐ type II, proximal myotonic myopathy, PROMM (*CNBP*)
- ☐ Spinal bulbar muscular atrophy, SBMA, Kennedy disease (*AR*)
- ☐ Spinal muscular atrophy type I / II / III (*SMN1*)

Nutrigenetics
Celiac disease → please see "Gastrointestinal diseases
- ☐ Fructose intolerance, hereditary (*ALDOB*)
- ☐ Lactose intolerance (-13910C>T polymorphism of *LCT*)

Pancreatic diseases
- ☐ Hereditary pancreatitis (*PRSS1, SPINK1, CFTR*)

Periodic fevers
- ☐ comprehensive analysis periodic fevers (*NLRP3, MEFV, MVK, TNFRSF1A*)
- ☐ Chronic neurologic cutaneous and articular syndrome, CINCA / neonatal onset multisystemic inflammatory disease, NOMID (*NLRP3*)
- ☐ Familial cold autoinflammatory syndrome, FCAS (*NLRP3*)
- ☐ Familial mediterranean fever, FMF (*MEFV*)
- ☐ Hyper IgD Syndrome, HIDS (*MVK*)
- ☐ Muckle-Wells syndrome (*NLRP3*)
- ☐ TNF receptor-associated periodic fever syndrome, TRAPS (*TNFRSF1A*)

Pharmacogenetics
- ☐ 5-Fluorouracil (5-FU)-toxicity (*DPYD*, variant c.1905+1G>A)
- ☐ Irinotecan toxicity (*UGT1A1*)
- ☐ Statin toxicity (*SLCO1B1*), haplotype SLCO1B1*5
- ☐ Thiopurin-S-Methyltransferase Deficiency (*TPMT*, c.238G>C, c.460G>A und c.719A>G)

Short stature
- ☐ Achondroplasia (*FGFR3*)
- ☐ Hypochondroplasia (*FGFR3*)
- ☐ SHOX deficiency (*SHOX*)

Silver-Russell syndrome, SRS → please see "Complex syndromes"
- ☐ Thanatophoric Dysplasia (*FGFR3*)

Thrombophilia / Atherosclerosis
- ☐ Angiotensin converting enzyme (*ACE*, del/ins polymorphism)
- ☐ Antithrombin (AT3) deficiency (*SERPINC1*)
- ☐ ß-Fibrinogen (*FGB*, -455G>A polymorphism)
- ☐ Factor V (*F5*)
  - ☐ Leiden mutation p.(Arg534Gln) ☐ HR2 haplotype p.(His1327Arg)
- ☐ Factor XIII, A1 subunit (*F13A1*, p.(Val35Leu))
- ☐ Glycoprotein Ia (*ITGA2*) c.759C>T (legacy c.807C>T)
- ☐ Glycoprotein IIIa (*ITGB3*) p.Leu59Pro (legacy L33P)
- ☐ Homocysteinemia (*MTHFR*) p.(Ala222Val), p.(Glu429Ala)
- ☐ Plasminogen activator inhibitor type1, PAI1 (*SERPINE1*, 4G/5G polymorphism)
- ☐ Prothrombin, FII (*F2*) mutation G20210A
- ☐ Protein C deficiency (*PROC*)
- ☐ Protein C receptor (*PROCR*)
- ☐ Protein Z deficiency (*PROZ*)
- ☐ Protein Z dependent protease inhibitor (*SERPINA10*)
- ☐ Thrombomodulin defect (*THBD*)

Uniparental disomy
☐ UPD2* ☐ UPD6* ☐ UPD7*
☐ UPD11* ☐ UPD14* ☐ UPD15*
*additionally, parents' blood samples required

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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.

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.

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

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

Using molecular genetic or molecular cytogenetic screening tests incidental findings not related to the primary clinical indication may be revealed.

☐ I want to be informed about clinically relevant incidental findings

☐ I refrain from being informed about diagnostically relevant incidental findings.

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