

AOK	LKK	BKK	IKK	VdAK	AEV	Knappschaft
Name des Versicherten						
geb. am						
Kassen-Nr.		Versicherungs-Nr.		Status		
Betriebsstätten-Nr.		Arzt-Nr.		Datum		

## Labor für Molekulare Genetik

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In Zusammenarbeit mit dem Ambulanzzentrum UKE GmbH

### Klinische Angaben:

### Einsender:

(Stempel)

### Hyperandrogenämie/Adrenogenitales Syndrom

- Steroid-21-Hydroxylase-Defizienz (CYP21A2-Gen, MIM +201910)
- Steroid-11- $\beta$ -Hydroxylase-Defizienz (CYP11B1-Gen, MIM 610613)
- Steroid-17- $\alpha$ -Hydroxylase-Defizienz (Cyp17-Gen, MIM \*609300)
- Steroid-3- $\beta$ -Dehydrogenase-Defizienz (3HSD-Gen, +MIM 201810)
- Cytochrom P450 Oxidoreductase (POR, MIM \*124015)

### Diabetes

- MODY Typ 1, Hepatic Nuclear Factor 4  $\alpha$  (HNF4 $\alpha$ -Gen, MIM \*600281)
- MODY Typ 2, Glucokinase (GCK-Gen, MIM \*138079)
- MODY Typ 3, Hepatic Nuclear Factor 1 $\alpha$  (HNF1 $\alpha$ /TCF1-Gen, MIM \*142410)
- MODY Typ 4, Homeodomain Transcriptionfaktor IPF (IPF-Gen, MIM 600733)
- MODY Typ 5, Renale Cysten und Diabetes (HNF1B-Gen, MIM \*189907)
- MODY Typ 6, Beta-cell E-box Transaktivator 2 (NEUROD-Gen, MIM 601724)
- Mitochondrial induzierte Diabetes (MTTL-Gen, MIM \*590050; MTTK-Gen, MIM \*590060; MTTS-Gen, MIM \*59085)

### Andrologie

- Azoospermie (AZF, MIM #415000; USP9Y-Gen, MIM \*400005)
- Cystische Fibrose, CBAVD (CFTR1-Gen, MIM \*602421)
- Polymerase-Gamma (POLG1-Gen, MIM \*174763)
- Ribonuklease L (RNAsel-Gen, MIM \*180435)

### Endokrinologie und Stoffwechsel

- Aarskog Syndrom, Faciogenital Dysplasia 1 (FGD1-Gen, MIM #305400)
- Acyl-CoA Dehydrogenase, long chain Defizienz (ACADL-Gen, MIM \*609676)
- Acyl-CoA Dehydrogenase, very long chain Defizienz (ACADVL-Gen, MIM \*609575)
- Alagille Syndrom (Jagged-1 Gene, JAG, AGS MIM +601920; NOTCH2-Gen, MIM \*600275)
- Albright-Syndrom (Guanin-Nucleotide-Binding-Proteine, GNAS-Gen, MIM +139320)
- Aldolase B Defizienz (Fruktose Intoleranz, ALDOB-Gen, MIM +229600)
- Alport Syndrom, X-linked (Col4A5-Gen, ATS, MIM \*303630)
- Alport Syndrom, autosomal rezessiv (Col4A3-Gen, MIM \*120070, Col4A4-Gen MIM \*120131)
- Angiotensin Converting Enzyme (ACE-Gen, I/D-Polymorphismus, MIM +106180)
- Apolipoprotein A1/CIII (Apolipoprotein-Cluster Chromosome 11, ApoA1/CIII-Gen, MIM \*107680 und MIM \*107720)
- Apolipoprotein AIV (ApoA4-Gen, MIM \*107690)
- Apolipoprotein AV (ApoA5-Gen, MIM \*606368)
- Apolipoprotein B (ApoB 100-Gen, Arg3500Gln, MIM +107730)
- Apolipoprotein E (ApoE, E2/E3/E4-Isoforms, MIM +107741)
- ATP-Bindungs Cassette Transporter A 1 (ABCA1-Gen, MIM +600046)
- Carnitin Defizienz (primary systemic, Solute Carrier Family 22 member 5, SLC22A5-Gen, MIM \*603377)
- Catechol-O-Methyltransferase (Catechol- und Östrogenkatabolismus, COMT-Gen, MIM +116790)
- Cholesterolester Transferprotein (Lipid-Transfer-Protein, CETP-Gen, MIM \*118470)
- 11- $\beta$ -Cortisol-Ketoreduktase-Defizienz (11- $\beta$ -Hydroxysteroid-Dehydrogenase, HSD11-Gen, MIM +218030)

- Diabetes insipidus (Neurohypophysial Type, Arginine Vasopressin, AVP-Gen, MIM \*192340)
- Ehlers-Danlos-Syndrome Typ VIIa (Collagen-1A1-Gen, COL1A1, MIM \*120150)
- Familiärer Hyperaldosteronämie (Glucocorticoid suppressable, Chimeric CYP11B1/CYP11B2, GRA, MIM #103900)
- Familiäre Hypercholesterinämie (LDL-Rezeptor-Gen, LDLR, MIM \*606945)
- Hypercholesterinämie, autosomal dominant (LDL-Rezeptor-Gen, MIM\*606945, ApoB, MIM \*107730, PCSK9-Gen +607786)
- Familiäre hypokalziurische Hyper- und Hypokalzämie (Calcium-Sensing-Rezeptor-Gen, CASR, MIM +601199)
- Familiäres Mittelmeerfieber (Recurrent Polyserositis, FMF, MEVF-Gen \*608107)
- Familiäre Schilddrüsenhormone Resistenz (Thyroid Hormone- $\beta$ -Rezeptor, THRB-Gen, MIM \*190160)
- Follikel-stimulierendes-Hormon FSH-Rezeptor (FSH-Rezeptor, FSHR-Gen, MIM \*136435)
- Galactosidase alpha Defizienz, Morbus Fabry (alpha Galactosidase, GLA-Gen, MIM \*300644)
- Gaucher Syndrom, Typ I, II and III (acid-beta glucosidase GBA-Gen, MIM \*606463)
- Glutarazidämie I (Glutaryl-CoA dehydrogenase, GCDH-Gen, MIM \*608801)
- Glykogen Speicher-Krankheit II (Morbus Pompe, saure alpha-1,4 Glucosidase, GAA-Gen, MIM \*606800)
- Hämochromatose (HFE-Gen, MIM +235200)
- HPT-Jaw Tumor Syndrome (Parafibromin-Gen, HPT2-Gen, MIM 145001)
- Hyperbilirubinämie, Gilbert Syndrome, Morbus Meulengracht (UGT1A1-Gen, MIM \*191740)
- Hyperlipoproteinämie Typ 1 (Lipoproteinlipase-Gen, LPL-Gen, MIM \*609708)
- Hypophosphatämie, Hereditär (Vitamine-D-resistente Rachitis, X-chromosomal, PHEX-Gen, MIM \*300550)
- Hypophosphatasie (Alkalische Phosphatase –Leber/Niere/Knochen, ALPL-Gen, MIM \*171760)
- Isovaleriansäure Azidämie (Isovaleryl CoA dehydrogenase Gen, IVD-Gen, MIM \*607036)
- KLOTHO (Susceptibility for CHD, Longevity, KL-Gen, MIM +604824)
- Laktose Intoleranz (Hypolaktasie, Latase, LCT-Gen, MIM \*603202)
- Matrix Metalloproteinase 3 (Human Fibroblast Stromelysin, MMP3-Gen, MIM \*185250)
- McCune-Albright Syndrom (Guanin-Nucleotide-Bindingprotein, GNAS-Gen, MIM +139320)
- Methymalonsäure Acidurie (Methymalonyl-CoA Mutase, MUT-Gen, MIM \*609058)
- Methymalonsäure Acidurie, cbIA Typ (MMAA-Gen, MIM \*607481)
- Methymalonic Aciduria, cbIB TYPE (MMAB-Gen, MIM \*607568)
- Multiple Acyl-CoA Dehydrogenase Defizienz (Glutarsäure Acidurie 2; ETFAGene, MIM \*608503; ETFB-Gen, MIM \*130410, ETFDH-Gen, MIM \*231675)
- Multiple Exostosen Typ 1 (Multiple Osteochondromatosis Type 1, Exostosin 1, EXT1, MIM \*608177)
- Multiple Exostosen Typ 2 (Exostosin 2-Gen, EXT2, MIM #133701)

- Niemann-Pick Krankheit Typ A** (Sphingomyelin phosphodiesterase-1-Gen (SMPD1, MIM \*607608)
- Niemann-Pick Krankheit Typ C1** (NPC1-Gen, MIM \*607623)
- Nitroxid Synthase Typ 3** (endothelial, eNOS-Gen, MIM +163729)
- Östrogen- $\alpha$ -Rezeptor** (Estrogen Receptor  $\alpha$ -Gen, ESR, MIM +133430)
- Östrogen- $\beta$ -Rezeptor** (Estrogen Receptor  $\beta$ -Gen, ESRB, MIM \*601663)
- Osteochondrodysplasia, X-chromosomal** (Spondyloepiphyseal Dysplasia tarda, SEDL, TRAPPC2-Gen, MIM \*300202)
- Osteogenesis Imperfecta Typ I und Typ IIa** (COL1A1-Gen, MIM \*120150, Col1A2-Gen, MIM \*120160)
- Osteogenesis Imperfecta Typ IIb und Typ VII** (CRTAP-Gen, MIM \*605497)
- Osteoporosis Pseudoglioma Syndrom** (OPP, LDL-Receptor Related Protein 5, LRP5, MIM \*603506)
- Paraoxonase** (PON1-Gen, MIM +168820)
- Pendred-Syndrom** (Hörstörung und Struma, Solute carrier Family 26 Member 4-Gen, SLC26A4, MIM \*605646)
- Peroxisome Proliferator activated Receptor  $\alpha$**  (PPARA-Gen, MIM +170998)
- Peroxisome Proliferator activated Receptor  $\gamma$**  (PPARG-Gen, MIM \*601487)
- Polyglanduläre endokrine Insuffizienz Typ I** (Autoimmune-Regulator-Gen, AIRE, MIM \*607358)
- Propionacädemie** (Propionyl-CoA Carboxylase Gen, PCCA-Gen, MIM \*232000; PCCB-Gen, MIM \*232050)
- Pseudoaldosteronismus, Liddle Syndrom** (SCNN1B-Gen, MIM \*600760; SCNN1G-Gen, MIM \*600761)
- Pseudohypoparathyreoidismus, Typ IA; PHP1A** (GNAS-Gen, MIM +139320)
- Pseudo-Vitamin-D-resistente Rachitis** (25-Hydroxy-Vitamine-D- $1\alpha$ -Hydroxylase-Defizienz, CYP27B1-Gen, MIM \*609506)
- Sichelzellanämie** (Hämoglobin Beta Locus, HBB-Gen, MIM +141900)
- Steroid-5 $\alpha$ -Reduktase2** (Männlicher Pseudohemaphroditismus, SRDA2-Gen, \*607306)
- Testikuläre Feminisierung, Androgen Resistenz** (Androgen Rezeptor Gen, AR, MIM \*313700)
- TSH-Resistenz** (Thyrotropin Rezeptor- Gen, TSHR, MIM +603372)
- $\beta$ -Thalassämie** (Hämoglobin- $\beta$ -Locus, HBB-Gen, MIM +141900)
- Tyrosinämie, Typ I** (Fumarylacetoacetate Hydrolase, FAH-Gen, MIM +276700)
- Vitamin-D-Resistenz** (Vitamin-D-Rezeptor, VDR-Gen, MIM \*601769)
- Variiegata Porphyria** (Protoporphyrinogen Oxidase, PPOX-Gen, MIM \*600923)
- Wolfram-Syndrom** (Wolframin-Gen, WFS1, MIM \*606201)

## Neurologie und Mitochondriopathie

- Alpers-Syndrom**, frühkindliche Hepatoenzephalopathie (POLG-Gen, MIM \*174763)
- MtDNA Depletion-Syndrom**, hepatocellulär Form (DGUOK-Gen, MIM \*601465, MPV17-Gen, MIM \*137960, C10ORF2-Gen, MIM \*606075)
- Chronisch progressive externe Ophthalmoplegie** (CPEO, mt-tRNA-Leu(UUR), MIM \*590050; mt-tRNA Ile, MIM \*590045; mt-tRNA Leu(CUN), MIM \*590055; mt-tRNA Ala, MIM \*590000; mt-tRNA Asn, MIM \*590010)
- Kearns-Sayre-Syndrom einschließlich Pearson Syndrome** (KSS, mt-DNA Deletionen, MTTL-Gen, MIM \*590050)
- Leber hereditäre Optikus Neuropathie** (LHON, MTND1-Gen, MIM \*516000; MTND2-Gen, MIM \*516001; MTND4-Gen, MIM 516004; MTND5-Gen, MIM \*516005; MTND6-Gen, MIM \*516005; MTCO1-Gen, MIM \*516030; MTCO3-Gen, MIM \*516050; MTATP6-Gen, \*MIM 516060; MTCYB-Gen, MIM \*516020)
- Leigh-Syndrom** (Necrotisierende Enzephalopathie, LS, MTND2, \*516001; MTND3-Gen, MIM \*516003; MTND5-Gen, MIM \*516005; MTND6-Gen, MIM \*516006; NDUfv1-Gen, MIM \*161015; NDUFS1-Gen, \*157655; NDUFS3-Gen, MIM \*603846; NDUFS4-Gen, MIM \*602694; NDUFS7-Gen, MIM 601825; NDUFS8-Gen, MIM \*602141; NDUFA2-Gen, MIM \*602137; C8ORT38-Gen, MIM \*61492; SURF1-Gen, MIM 185620; MTATP6-Synthase-Gen, MIM 516060; MTTV-Gen, MIM \*590105; MTTK-Gen, MIM \*590060; MTTW-Gen, MIM \*590095; MTTL1-Gen, MIM \*590050)
- Musculäre Dystrophie Duchenne/Becker** (Dystrophin-Gen, MIM 310200)
- Mitochondriale Enzephalomyelopathie, Laktat Azidose, schlaganfallähnliche Symptome, MELAS** (MTTL1-Gen, MIM \*590050; MTTQ-Gen, MIM \*590030; MTTH-Gen, MIM \*590040; MTTK-Gen, MIM \*590060, MTTs1-Gen, MIM \*590080; MTND1-Gen, MIM 516000; MTND5-Gen, MIM \*515005; MTND6-Gen, MIM \*516006; MTTs2-Gen, MIM \*590085)
- Mitochondriale Enzephalomyopathie mit „Ragged Red Fibers**, MERRF, (MTTL1-Gen, MTTH-Gen, MIM \*590040; MTTK-Gen, MIM \*590060, MTTs1-Gen, MIM \*590080; MTND5-Gen, MIM \*515005; MTTs2-Gen, MIM \*590085; MTTF-Gen, MIM \*590070)
- Neuropathie, Ataxie und Retinitis Pigmentosa, NARP** (MTATP6-Gen, MIM \*516060)
- Hereditäre Neuropathie mit Neigung zu Drucklähmungen, Tomakulöse Neuropathie, HNPP** (Peripheral Myelin Protein 22, PMP22-Gen, MIM \*601097)
- Pearson Syndrom, Sideroblastische Anämie, Marrow-Pancreas Syndrom**, (MTND4-Gen, MIM \*516003; MTND5-Gen, MIM \*516005)
- Spinale und bulbäre muskuläre Atrophie, SBMA, SMAX1** (Kennedy's Disease, CAG-Repeat, Androgen-Receptor-Gen, AR-Gen, MIM \*313700 )

## Onkologie

- Adenomatöse Polyposis Coli APC** (APC-Gen, MIM \*175100)
- Brustkrebs, Familiär** (BRCA1-Gen, MIM \*113705; BRCA2-Gen, +600185)
- Carney-Komplex Typ 1** (PRKAR1A-Gen, MIM \*188830)
- Cowden Syndrom** (PTEN-Gen, MIM \*601728)

- Familiärer isolierter Hyperparathyreoidismus, FIHP** (HRPT2-Gen, MIM \*602393; MEN1-Gen, MIM \*131100; HRPT3, MIM %610071 , CASR-Gen, MIM +601199)
- Familiäre isolierte Somatopinome (FIS)**, GH sezernierende Hypophysendome (Aryl Hydrocarbon Receptor Interacting Protein, AIP-Gen, MIM \*605555)
- Familiäre isolierte Pituitary Adenoma (FIPA)**; (Aryl Hydrocarbon Receptor Interacting Protein, AIP-Gen, MIM \*605555)
- Hereditäres non-polyposis Colon-Carcinom, Lynch-Syndrom** (HNPCC, MSH2-Gen, MIM \*609309; MLH1-Gen, MIM \*120436; MSH6-Gen, MIM \*600678; MLH3-Gen, \*604395; PMS1-Gen, MIM \*600258; PMS2-Gen, MIM \*600259; TGFBR2-Gen, MIM \*190182)
- HPT-Jaw Tumor Syndrom** (HRTP2, Parafibrinogen-Gen, MIM \*607393)
- Li Fraumeni-Syndrom** (Tumorsuppressor-Gen p53, TP53, MIM \*191170)
- Mikrosatelliten Instabilität, MSI**
- Multiple endokrine Neoplasie Typ 1** (MEN 1-Gen, MIM \*131100)
- Multiple endokrine Neoplasie Typ 2A and 2B** (RET-Proto-Onkogen, MIM +164761)
- Multiple endokrine Neoplasie Typ 4** (CDKN1B-Gen, MIM \*600778)
- Multiple Phäochromozytome, Paragangliom** (PGL1, Succinat Dehydrogenase Subunit D, SDHD-Gen, MIM \*602690; PGL3, SDH Subunit C, SDHC-Gen, MIM \*602413; PGL4, SDH Subunit B, SDHB-Gen, MIM \*185470)
- Neurofibromatose Typ 1** (Neurofibrinogen-Gen, NF1-Gen, MIM +162200)
- Septin9** (DNA-Methylierung, MIM \*604061)
- Von Hippel-Lindau-Syndrom** (VHL-Gen, MIM \*608537)

## Thrombophilie/Hämophilie

- Antithrombin III-Defizienz** (Antithrombin-Gen, MIM +107300)
- Faktor V Leiden, APC-Resistenz** (Faktor V-Gen, G1691A, MIM \*612309)
- Hemophilia A** (Faktor VIII-Gen, F8-Gen, MIM +306700)
- Hemophilia B** (Faktor IX, F9-Gen, MIM \*300746)
- Integrin, Platelet Glycoprotein IIIa** (ITGB3-Gen, MIM +173470)
- Methylentetrahydrofolat-Reduktase** (MTHFR-Gen, Ala677Val, MIM \*607093)
- Plasminogenaktivator Inhibitor 1** (Serinproteinase-Inhibitor 1, PAI1-Gen, MIM \*173360)
- Protein C-Defizienz** (Protein C-Gen, MIM \*612283)
- Protein S-Defizienz** (Protein S-alpha-Gen, PROS1, MIM \*176880)
- Prothrombin – Faktor 2** (Factor 2-Gen, G20210A, MIM +176930)

## Immunologie

- HIV-Suszeptibilität** (CC Chemokine Receptor, CCR5, MIM \*601373)
- Interleukine 1a** (IL1a-Gen, MIM \*147760)
- Interleukine 1b** (IL1b-Gen, MIM \*147720)
- Interleukine 6** (Interferon- $\beta$ -2-Gen, IL6, MIM \*147620)
- Interleukine 10** (Cytokine Synthesis Inhibitory Factor, IL10-Gen, MIM \*124092)
- Transforming Growth Factor b 1** (TGFB1-Gen, MIM \*190180)
- Transforming Growth Factor b 1-Rezeptor** (TGFB1-Gen, MIM \*190181)
- Tumor-Necrosis-Factor-a** (TNFa-Gen, MIM \*191160)
- Wiskott-Aldrich Syndrome** (WAS-Gen, MIM \*300392)

## Pharmakogenetik

- Cytochrom P450 1A1** (Aryl Kohlenwasserstoff-Hydroxylase, Cyp1A1-Gen, MIM \*108330)
- Cytochrom P450 1A2** (Aromatic compound inducible, Cyp1A2-Gen, MIM +124060)
- Cytochrom P450 1B1** (Microsomal Monooxidase, Cyp1B1-Gen, MIM \*601771)
- Cytochrom P450 2A6** (Coumarin 7-Hydroxylase, Cyp2A6-Gen, MIM +122720)
- Cytochrom P450 2C9** (Warfarin/Tolbutamid-Hydroxylase, Cyp2C9-Gen, MIM \*601130)
- Cytochrom P450 2C19** (Mephenytoin 4-Hydroxylase, Cyp2C19-Gen, MIM \*124020)
- Cytochrom P450 2D6** (Debrisoquine 4-Hydroxylase, Cyp2D6-Gen, MIM +124030)
- Cytochrom P450 2E1** (Ethanol inducible P450, Cyp2E1-Gen, MIM \*124040)
- Cytochrom P450 3A4** (Glucocorticoid inducible P450, Nifedipin Hydroxylase, Cyp3A4-Gen, MIM \*124010)
- Dihydropyrimidin Dehydrogenase** (Fluoruracil-Toxicity, DPYD-Gen, MIM +274270)
- Epoxid Hydroxylase** (Microsomal, EPHX-Gen, MIM 132810)
- Glutathion-S-Transferase M1** (Liver- and Fibroblast-specific, GSTM1-Gen, MIM 138350)
- Glutathion-S-Transferase M3** (Brain-specific, GSTM3-Gen, MIM 138390)
- Glutathion-S-Transferase T1** (Liver- and Erythrocyte-specific, GSTT1-Gen, MIM \*600436)
- Glutathion-S-Transferase P1** (Fatty acid ethyl ester synthase III, GSTP1-Gen, MIM \*134660)
- N-Acetyl-Transferase Typ1** (Arylamine N-Acetyl transferase Type1, NAT1-Gen, MIM \*108345)
- N-Acetyl-Transferase Typ2** (Arylamine N-Acetyl transferase Type2, NAT2-Gen, MIM \*612182)
- Sulfotransferase 1A1** (Phenol-, SULT1A1Gen, MIM \*171150)
- Sulfotransferase 1C1** (Hydroxylamin, SULT1C1Gen, MIM \*602385)
- Superoxid Dismutase Typ 2** (Mn-abhängig, SOD2, MIM \*147460)
- Superoxid Dismutase Typ 3** (SOD3, MIM \*185490)
- Thiopurin-S-Methyltransferase** (Mercaptopurin-Toxicity, TPMT, MIM \*187680)

Andere: \_\_\_\_\_