| AOK | LKK | BKK | IKK | VdAK | AEV | Knappschaft |
|---|---|---|---|---|---|---|
| | | | | | | |

Name des Versicherten

geb. am

| Kassen-Nr. | Versicherungs-Nr. | Status |
|---|---|---|
| | | |

| Betriebsstätten-Nr. | Arzt-Nr. | Datum |
|---|---|---|
| | | |

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In Zusammenarbeit mit dem Ambulanzzentrum UKE GmbH


**Einsender:**

**(Stempel)**

## Hyperandrogenämie/Adrenogenitales Syndrom

- ☐ **Steroid-21-Hydroxylase-Defizienz** (CYP21A2-Gen, MIM +201910)
- ☐ **Steroid-11-β-Hydroxylase- Defizienz** (CYP11B1-Gen, MIM 610613)
- ☐ **Steroid-17-α-Hydroxylase- Defizienz** (Cyp17-Gen, MIM *609300)
- ☐ **Steroid–3-β-Dehydrogenase- Defizienz** (3HSD-Gen, +MIM 201810)
- ☐ **Cytochrom P450 Oxidoreductase** (POR, MIM *124015)

## Diabetes

- ☐ **MODY Typ 1,** Hepatic Nuclear Factor 4 α (HNF4α-Gen, MIM *600281)
- ☐ **MODY Typ 2,** Glucokinase (GCK-Gen, MIM *138079)
- ☐ **MODY Typ 3,** Hepatic Nuclear Factor 1α (HNF1α/TCF1-Gen, MIM *142410)
- ☐ **MODY Typ 4,** Homeodomain Transcriptionfactor IPF (IPF-Gen, MIM 600733)
- ☐ **MODY Typ 5**, Renale Cysten und Diabetes (HNF1B-Gen, MIM *189907)
- ☐ **MODY Typ 6,** Beta-cell E-box Transactivator 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)
- ☐ **Apolipoporotein AI/CIII** (Apolipoprotein-Cluster Chrosome 11, ApoA1/CIIIGene, 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 Östrogenkatabolism, COMT-Gen, MIM +116790)
- ☐ **Cholesterolester Transferprotein** (Lipid-Transfer-Protein, CETP-Gen, MIM *118470)
- ☐ **11-β-Cortisol-Ketoreduktase-Defizienz** (11-β-Hydroxysteroid-Dehydrogenase, HSD11-Gen, MIM +218030)
- ☐ **Diabetes insipidus** (Neurohypophyseal 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- and Hypokalzämie** (Calcium-Sensing-Receptor-Gen, CASR, MIM +601199)
- ☐ **Familiäres Mittelmeerfieber** (Recurrent Polyserositis, FMF, MEVF-Gen *608107)
- ☐ **Familiäre Schilddrüsenhormone Resistenz** (Thyroid Hormone-β-Receptor, THRB-Gen, MIM *190160)
- ☐ **Follikel-stimulierendes-Hormon FSH-Rezeptor** (FSH-Receptor, 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, HRTP2-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)
- ☐ **Hyppophosphatasie** (Alkalische Phosphatase –Leber/Niere/Knochen, ALPL-Gen, MIM *171760)
- ☐ **Isoveleriansäure Azädimie** (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, cblA Typ** (MMAA-Gen, MIM *607481)
- ☐ **Methymalonic Aciduria, cblB TYPE** (MMAB-Gen, MIM *607568 )
- ☐ **Multiple Acyl-CoA Dehydrogenase Defizienz** (Glutarsäure Acidurie 2; ETFAGene, MIM *608503; ETFB-Gen, MIM *130410, ETFDH-Gene, 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-α-Rezeptor** (Estrogen Receptor α-Gen, ESRA, MIM +133430)
- ☐ **Östrogen-β-Rezeptor** (Estrogen Receptor β-Gen, ESRB, MIM *601663)
- ☐ **Osteochondrodysplasia, X-chromosomal** (Spondyloepiphysal 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, LPR5, 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 a** (PPARA-Gen, MIM +170998)
- ☐ **Peroxisome Proliferator activated Receptor g** (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-Gene MIM *600760; SCNN1G-Gene MIM *600761)
- ☐ **Pseudohypoparathyreoidismus, Typ IA; PHP1A** (GNAS-Gen, MIM +139320)
- ☐ **Pseudo-Vitamin-D-resistente Rachitis** (25-Hydroxy-Vitamine-D-α-Hydroxylase-Defizienz, CYP27B1-Gen, MIM *609506)
- ☐ **Sichelzellanämie** (Hämoglobin Beta Locus, HBB-Gen MIM +141900)
- ☐ **Steroid-5α-Reduktase2** (Männlicher Pseudohermaphroditismus, SRDA2-Gen, *607306)
- ☐ **Testikuläre Feminisierung, Androgen Resistenz** (Androgen Rezeptor Gen, AR, MIM *313700)
- ☐ **TSH-Resistenz** (Thyrotropin Rezeptor- Gen, TSHR, MIM +603372)
- ☐ **β-Thalassämie** (Hemoglobin-ß-Locus, HBB-Gen, MIM +141900)
- ☐ **Tyrosinämie, Typ I** (Fumarylacetoacetate Hydrolase, FAH-Gen, MIM +276700)
- ☐ **Vitamin-D-Resistenz** (Vitamin-D-Rezeptor, VDR-Gen, MIM *601769)
- ☐ **Variegata Porphyria** (Protoporphyrinogen Oxidase, PPOX-Gen, MIM *600923)
- ☐ **Wolfram-Syndrom** (Wolframin-Gene, WFS1, MIM *606201)

## Neurologie und Mitochondriopathie

- ☐ **Alpers-Syndrom**, frühkindliche Hepatoenzephalopathie (POLG-Gen, MIM *174763)
- ☐ **MtDNA Depletion-Syndrom**, hepatocellebrär Form (DGUOK-Gen, MIM *601465, MPV17-Gen, MIM *137960, C10ORF2-Gen, MIM *606075)
- ☐ **Chronisch progressive externe Ophtalmoplegie** (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 Encephalopathie, LS, MTND2, *516001; MTND3-Gen, MIM *516003; MTND5-Gen, MIM *516005; MTND6-Gen, MIM *516006; NDUfV1-Gen, MIM *161015; NDUFS1-Gen, *157655.Gen; NDUFS3-Gen, MIM *603846; NDUFS4-Gen, MIM *602694; NDUIFS7-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 and bulbäre muskuläre Atrophie**, SBMA, SMAX1 (Kennedy's Disese, 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 Hypophysendnome (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, Parafibromin-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** (Neurofibromin-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)
- ☐ **Factor 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)
- ☐ **Methylentetrahydrofolate-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-Gene, PROS1, MIM *176880)
- ☐ **Prothrombin – Faktor 2** (Factor 2-Gene, G20210A, MIM +176930)

## Immunologie

- ☐ **HIV-Susceptibilitaty** (CC Chemokine Receptor, CCR5, MIM *601373)
- ☐ **Interleukine 1a** (IL1a-Gen, MIM *147760)
- ☐ **Interleukine 1b** (IL1b-Gen, MIM *147720)
- ☐ **Interleukine 6** (Interferone-b-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−Receptor** (TGFBR1-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:**_____