BIOGLOBE
The home of life science

Patient:					
Name:					
Date of birth:					
Gender:	Female			Male	
Address:					

Address of Referring Doctor:

Phone:
E-mail:

Anamnesis:

Family history:

Hyperandrogenemia/Congenital Adrenal Hyperplasia/ Pseudo-hermaphroditism

- ☐ **Aromatase-Deficiency** (CYP19, MIM 107910)
- ☐ **Steroid-21-Hydroxylase-Deficiency** (CYP21B, MIM 201910)
- ☐ **Steroid-11-β-Hydroxylase- Deficiency** (CYP11B1, MIM 202010)
- ☐ **Steroid-17-α-Hydroxylase- Deficiency** (Cyp17, MIM 202110)
- ☐ **Steroid–3-β-Dehydrogenase- Deficiency** (3HSD, MIM 201810)

Diabetes

- ☐ **MODY Type 2,** Glucokinase (GCK, MIM 138079)
- ☐ **MODY Type 4,** Homeodomain Transcriptionfactor IPF (IPF, MIM 600733)
- ☐ **MODY Type 6,** Beta-cell E-box Transactivator 2 (NEUROD, MIM 601724)
- ☐ **Mitochondrial induced Diabetes** (mt-tRNA Leu(UUR), MIM 590050; mt-tRNA Lys, MIM 590060; mt-tRNASer, MIM 59085)

Andrology

- ☐ **Azoospermiafactor Yq11** (AZF, MIM 415000)
- ☐ **Cystic Fibrosis, CBAVD** (CFTR1, MIM 602421)
- ☐ **Polymerase-Gamma** (POLG1, MIM 174763)
- ☐ **Ribonuclease L** (RNAsel, OMIM 180435)

Neurology und Mitochondriopathies

- ☐ **Chronic progressive external Ophtalmoplegia** (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-Syndrome including Pearson Syndrome** (KSS, mt-DNA Deletionen, mt-tRNA-Leu(UUR), MIM 590050)
- ☐ **Leber hereditary Opticus Neuropathy** (LHON, ND1, MIM 516000; ND2, MIM 516001; mt-CO1, MIM 516030; mt-ATP6, MIM 516060; mt-CO3, MIM 516050; ND4L, MIM 516004; ND5, MIM 516005; ND6, MIM 516006; mt-CYB, MIM 516020)
- ☐ **Leigh-Syndrome** (Necrotizing encephalopathy, LS, SURF1, MIM 185620; ATP6-Synthase, MIM 516060; mt-tRNA(Val), MIM 590105)
- ☐ **Muscular Dystrophy Duchenne/Becker** (Dystrophin-Gene, MIM 310200)
- ☐ **Myoclonic Epilepsy, Stroke like Episodes, Lactat acidosis** (MELAS, mt-tRNA Leu (UUR) MIM 590050; mt-tRNA Val, MIM 590105; mt-tRNA Val, 590105; mt-tRNA Phe, MIM 590070; ND1, MIM 516000; ND2, MIM 515001; ND4, MIM 515003; ND5, MIM 516004; mt-CO3, MIM 516050; mt-CYB, MIM 516020)
- ☐ **Myoclonic Epilepsy, red ragged fibers** (MERRF, mt-tRNA Lys, MIM 590060)
- ☐ **Neuropathy, Ataxia und Retinitis Pigmentosa** (NARP, mt-ATP6, MIM 516060)
- ☐ **Pearson Syndrome** (Sideroblastic anemia, Marrow-Pancreas Syndrome, mtND4, MIM 516003; mtND5 516005)
- ☐ **Spinal and Bulbar Muscular Atrophy SMAX1** (Kennedy's Disease, Androgen-Receptor-Gene, CAG-Repeat, SMA, MIM313200)

Oncology

- ☐ **Adenomatous Polyposis of the Colon**, APC (APC-Gene, MIM 175100)
- ☐ **Hereditary non-polyposis Colon-Carcinoma**, HNPCC (Microsatellite-Instability)
- ☐ **HPT-Jaw Tumor Syndrome** (HRTP2, Parafibromin-Gene, MIM 145001)
- ☐ **Li Fraumeni-Syndrome** (Tumorsuppressor-Gene p53, TP53, MIM191170)
- ☐ **Multiple endokrine Neoplasia Type 1** (MEN 1-Gene, MIM 131100)
- ☐ **Multiple endokrine Neoplasia Type 2** (RET-Proto-Onkogene, MIM 171400)
- ☐ **Multiple Pheochromocytoma, Paraganglioma** (Succinat Dehydrogenase Subunit D, SDHD-Gene, MIM 602690;SDH Subunit B, SDHB-Gene, MIM 185470, SDH Subunit C, SDHC-Gene, MIM 602413)
- ☐ **Neurofibromatosis Type 1** (Neurofibromin-Gene, NF1, MIM 162200)
- ☐ **Peutz-Jeghers Syndrome** (Hamartomatous intestinal polyposis, Serine/Threonine Proteinkinase 11-Gene, PJS, MIM602216)
- ☐ **Von Hippel-Lindau-Syndrome** (VHL-Gene, MIM 193300)

Thrombophilia

- ☐ **Antithrombin III-Deficiency** (Antithrombin-Gene, MIM 107300)
- ☐ **Factor V Leiden, APC-Resistance** (Factor V-Gene, G1691A, MIM 227400)
- ☐ **Integrin, Platelet Glycoprotein IIIa** (ITGB3-Gene, MIM 173470)
- ☐ **Methylentetrahydrofolate-Reductase** (MTHFR-Gene, Ala677Val, MIM 236250)
- ☐ **Plasminogenactivator Inhibitor 1** (Serinproteinase-Inhibitor 1, PAI1, MIM 173360)
- ☐ **Protein C-Deficiency** (Protein C-Gen, MIM 176860)
- ☐ **Protein S-Deficiency** (Protein S-alpha-Gene, PROS1, MIM 176880)
- ☐ **Prothrombin – Factor 2** (Factor 2-Gene, G20210A, MIM 176930)

Endocrinology and Metabolism

- ☐ **Acyl-CoA Dehydrogenase, long chain deficiency** (ACADL, MIM 609676)
- ☐ **Acyl-CoA Dehydrogenase, very long chain deficiency** (ACADVL, MIM 609575)
- ☐ **Alagille Syndrome** (Jagged-1 Gene, JAG, AGS MIM 118450)
- ☐ **Albright-Syndrome** (Guanin-Nucleotide-Binding-Proteine, GNAS. MIM 139320)
- ☐ **Aldolase B Deficiency** (Fructose Intolerance, ALDOB, MIM 229600)
- ☐ **Alport Syndrome**, X-linked (Col4A5-Gene, ATS , MIM 301050)
- ☐ **Angiotensin Converting Enzyme** (ACE, I/D-Polymorphism, MIM 106180)
- ☐ **Apolipoporotein AI/CIII** (Apolipoprotein-Cluster Chrosome 11, ApoA1/CIII, MIM 107680 und 107720)
- ☐ **Apolipoprotein AIV** (ApoA4, MIM 107690)
- ☐ **Apolipoprotein AV** (ApoA5, MIM 606368)
- ☐ **Apolipoprotein B** (ApoB 100, Arg3500Gln, MIM 107730)
- ☐ **Apolipoprotein E** (ApoE, E2/E3/E4-Isoforms, MIM 107741)
- ☐ **ATP-Binding Cassette Transporter A 1** (ABCA1-Gen, MIM 600046)
- ☐ **Carnitine Deficiency** (primary systemic, Solute Carrier Family 22 member 5, SLC22A5, MIM 603377)
- ☐ **Catechol-O-Methyltransferase** (Catechol- und Estrogencatabolism, COMT, MIM 116790)

- ☐ **Cholesterolester Transferprotein** (Lipid-Transfer-Protein, CETP, MIM 118470)
- ☐ **11-β-Cortisol-Ketoreductase-Deficiency** (11-β-Hydroxysteroid-Dehydrogenase,HSD11, MIM 218030)
- ☐ **Diabetes insipidus** (Neurohypophyseal Type, Arginine Vasopressin-Gene, AVP, MIM 192340)
- ☐ **Ehlers-Danlos-Syndrome Typ VIIa** (Collagen-1A1-Gen, COL1A1, MIM 120150)
- ☐ **Estrogen-α-Receptor** (Estrogen Receptor α-Gene, ESRA, MIM 133430)
- ☐ **Estrogen-β-Receptor** (Estrogen Receptor β-Gene, ESRB, MIM 601663)
- ☐ **Faciogenital Dysplasia 1** (Aarskog Syndrome, FGD1, MIM 305400)
- ☐ **Familial Hyperaldosteronism**(Glucocorticoid suppressable, Chimeric CYP11B1/CYP11B2, GRA, MIM202010)
- ☐ **Familial Hypercholesterinemia** (LDL-Receptor-Gene, MIM 606945; Neural-Apoptosis-Gene, NARC1-Gen, 607786)
- ☐ **Familial hypocalciuric Hyper- and Hypocalcemia** (Calcium-Sensing- Receptor-Gene, CASR, MIM 601199)
- ☐ **Familial Mediterranian Fever** (Recurrent Polyserositis, FMF, MIM 249100)
- ☐ **Familial Thyroid Hormone Resistance** (Thyroid Hormone-β-Receptor, THRB, MIM 190160)
- ☐ **Follicle-Stimulating Hormone Receptor** (FSH-Receptor, FSHR, MIM 136435)
- ☐ **Gaucher Disease, Type I, II and III** (acid-beta glucosidase GBA, MIM 606463)
- ☐ **Glutaric Acidemia I** (Glutaryl-CoA dehydrogenase, GDH, MIM 608801)
- ☐ **Glycogen Storage Disease II** (Pompe disease, acid alpha-1,4-glucosidase, (GAA, MIM 600800)
- ☐ **Hemochromatosis** (HFE-Gene, MIM 235200)
- ☐ **High Density Bone Mass** (HBM, LDL-Receptor Related Protein 5, LPR5, MIM 603506)
- ☐ **HPT-Jaw Tumor Syndrome** (HRTP2, Parafibromin-Gene, MIM 145001)
- ☐ **Hyperbilirubinemia, Gilbert Syndrome** (UGT1A1-Gene MIM 191740)
- ☐ **Hyperlipoproteinemia Typ 1** (Lipoproteinlipase-Gene, LPL, MIM 238600)
- ☐ **Hypophosphatemia, Hereditary Type 1 and 2** (Vitamine-D-resistant Ricketts, X-chromosomal, Hyp1 und Hyp2, MIM 307800)
- ☐ **Hypophosphatasia** (Alcaline Phosphatase –Liver/Kidney/Bone, ALPL, MIM171760)
- ☐ **Isovaleric Acidemia** (Isovaleryl CoA dehydrogenase gene, IVD, MIM 607036)
- ☐ **KLOTHO** (Susceptibility for CHD, Longevity, KL, MIM 604824)
- ☐ **Lactose Intolerance** (Hypolactasia, Lactase, LCT, MIM 603202)
- ☐ **Matrix Metalloproteinase 3** (Human Fibroblast Stromelysin, MMP3, MIM 185250)
- ☐ **McCune-Albright Syndrom Syndrome** (Guanin-Nucleotide-Bindingprotein, GNAS, MIM 139320)
- ☐ **Methymalonic Aciduria** (Methymalonyl-CoA mutase, MUT, MIM 609058)
- ☐ **Methymalonic Aciduria, cblA TYPE** (MMAA-Gene, MMA, MIM 602481)
- ☐ **Methymalonic Aciduria, cblB TYPE** (MMAB-Gene, MMA, MIM 251110)
- ☐ **Multiple Acyl-CoA Dehydrogenase Deficiency** (glutaric aciduria 2; ETFA-gene, MIM 608503; ETFB-gene, MIM 130410, or ETFDH-gene, MIM 231675)
- ☐ **Multiple Exostosis Type 1** (Multiple Osteochondromatosis Type 1,Exostosin 1, EXT1, MIM 133700)
- ☐ **Multiple Exostosis Type 2** (Exostosin 2, EXT2, MIM 133701)
- ☐ **Niemann-Pick Disease, Type A** (Sphingomyelin phosphodiesterase-1 gene (SMPD1, MIM 607608)
- ☐ **Niemann-Pick Disease, Type C1** (NPC1-gene, MIM 607623)
- ☐ **Nitroxid Syntase Typ 3** (endothelial, eNOS, MIM163729)
- ☐ **Osteogenesis Imperfecta Typ I und Typ III** (Collagen-1A1-Gene, COL1A1, MIM 120150)
- ☐ **Osteoporosis Pseudoglioma Syndrome** (OPP, LDL-Receptor Related Protein 5, LPR5, MIM 603506)
- ☐ **Paraoxonase** (PON1, MIM 168820)
- ☐ **Pendred-Syndrome** (Deafness and Goiter, Solute carrier Family 26 Member 4, SLC26A4, MIM 605646)
- ☐ **Peroxisome Proliferator activated Receptor α** (PPARA, MIM 170998)
- ☐ **Peroxisome Proliferator activated Receptor χ** (PPARG, MIM 601487)
- ☐ **Polyglandular endocrine Insufficiency Type I** (Autoimmune-Regulator-Gene, AIRE, MIM 240300)
- ☐ **Propionic acidemia** (propionyl-CoA carboxylase, PCCA, MIM 232000, or PCCB, MIM 232050)
- ☐ **Pseudoaldosteronismus, Liddle Syndrome** (SCNN1B-Gene MIM 600760; SCNN1G-Gene600761)
- ☐ **Pseudo-Vitamine-D-resistant Ricketts** (25-Hydroxy-Vitamine-D-α-Hydroxylase-Deficiency, CYP27B1, MIM 264700)
- ☐ **Spondyloepiphysal Dysplasia tarda** (Osteochondrodysplasia, X-chromosomal, SEDL, MIM 300202)
- ☐ **Steroid-5α-Reductase2** (Male Pseudohermaphroditism,SRDA2, 607306)
- ☐ **Testicular Feminisation, Androgen Resistance** (Androgen Receptor Gene,AR, MIM 313700)
- ☐ **TSH-Resistance** (Thyrotropin receptor, TSHR, MIM 603372)
- ☐ **β-Thalassemia** (Hemoglobin-ß-Locus, MIM 141900)
- ☐ **Tyrosinemia, Type I** (Fumarylacetoacetate hydrolase, FAH, MIM 276700)
- ☐ **Vitamin-D-Resistance** (Vitamin-D-Receptor-Gene, VDR, MIM (601769)
- ☐ **Variegata Porphyria** (Protoporphyrinogen Oxidase, PPOX, MIM 600923)
- ☐ **Wolfram-Syndrome** (Wolframin-Gene, MIM 606201)

Immunology

- ☐ **HIV-Susceptibilitaty** (CC Chemokine Receptor, CCR5, MIM 601373)
- ☐ **Interleukine 1a** (IL1a-Gene, MIM 147760)
- ☐ **Interleukine 1b** (IL1b-Gene, MIM 147720)
- ☐ **Interleukine 6** (Interferone-β-2-Gene, IL6, MIM 147620)
- ☐ **Interleukine 10** (Cytokine Synthesis Inhibitory Factor, IL10-Gen, MIM 124092)
- ☐ **Transforming Growth Factor β 1** (TGFB1-Gene, MIM 190180)
- ☐ **Transforming Growth Factor β 1–Receptor** (TGFBR1-Gene, MIM 190181)
- ☐ **Tumor-Necrosis-Factor-α** (TNFα-Gene, MIM 191160)
- ☐ **Wiskott-Aldrich Syndrome** (WAS-Gene, MIM 30100)

Detoxification and Drug Metabolism

- ☐ **Cytochrome P450 1A1** (Aryl Hydrocarbone Hydroxylase, Cyp1A1, MIM108330)
- ☐ **Cytochrome P450 1A2** (Aromatic compound inducible, Cyp 1A2, MIM 124060)
- ☐ **Cytochrome P450 1B1** (Microsomal Monooxidase, Cyp1B1, MIM 601771)
- ☐ **Cytochrome P450 2A6** (Coumarin 7-Hydroxylase, Cyp2A6, MIM 122720)
- ☐ **Cytochrome P450 2C9** (Warfarin/Tolbutamid-Hydroxylase, Cyp2C9 MIM 601130)
- ☐ **Cytochrome P450 2C19** (Mephenytoin 4-Hydroxylase, Cyp2C19, MIM 124020)
- ☐ **Cytochrome P450 2D6** (Debrisoquine 4-Hydroxylase, Cyp2D6, MIM 124030)
- ☐ **Cytochrome P450 2E1** (Ethanol inducible P450, Cyp2E1, MIM 124040)
- ☐ **Cytochrome P450 3A4** (Glucocorticoid inducible P450, Nifedipin Hydroxylase, Cyp3A4 MIM 124010)
- ☐ **Dihydropyrimidin Dehydrogenase** (Fluoruracil-Toxicity, DPD, MIM 274270)
- ☐ **Epoxide Hydroxylase** (Microsomal, EPHX, MIM 132810)
- ☐ **Glutathion-S-Transferase M1** (Liver- and Fibroblast-specific, GSTM1, MIM 138350)
- ☐ **Glutathion-S-Transferase T1** (Liver- and Erythrocyte-specific, GSTT1, MIM 600436)
- ☐ **Glutathion-S-Transferase P1** (Fatty acid ethyl ester synthase III, GSTP1, MIM 134660)
- ☐ **N-Acetyl-Transferase Typ1** (Arylamine N-Acetyl transferase Type1, NAT1, MIM 243400)
- ☐ **N-Acetyl-Transferase Typ2** (Arylamine N-Acetyl transferase Type2, NAT2, MIM 243400)
- ☐ **Sulfotransferase 1A1** (Phenol-, SULT1A1, MIM 171150)
- ☐ **Sulfotransferase 1C1** (Hydroxylamine, SULT1C1, MIM 602385)
- ☐ **Superoxid Dismutase Typ 2** (Mn-dependent, SOD2, MIM 147460)
- ☐ **Superoxid Dismutase Typ 3** (SOD3, MIM 185490)
- ☐ **Thiopurine-S-Methyltransferase** (Mercaptopurin-Toxicity, TPMT, MIM 187680)

☐ **Other:**.....

Medical Genetics Services

The Molecular Genetic Laboratory of Bioglobe is a leader in the field of molecular diagnostics. Integrated in a network of co-operations with physicians, we offer a large number of DNA-oriented tests for many medical fields.
The highest priorities of Bioglobe are a high scientific standard, reliability and accuracy combined with competitive services. Our highly versatile laboratory and its personnel enable us to fulfil a variety of mandates tailored to our customers' needs. Customers who use our services have access to leading-edge technology and highly qualified personnel.

Sample shipment

For genetic tests 3-5 ml EDTA-blood is required. Please use the service of one of the world wide logistic companies (e.g. DHL). The material is stable for several days at ambient temperature. Cooling or freezing is not necessary. Do not forget to label the tubes with the patients name and date of birth.

Prices

The price of a test may vary significantly depending on the clinical symptoms of the patient or the family history. In some cases one or more genes need to be sequenced completely, in other case just one position in a gene has to be looked at. Please inquire for the price for your patient.

Turnaround times

For most tests no more than 3-4 weeks are needed after arrival of the sample in our lab.