



Requesting physician:

Patient information:

Request Form Metabolic disorders

Cause of examination: diagnostic predictive for familial mutation _____

Clinical information:

Family history:

(Check all that apply)

- Patient clinically affected
- No previous molecular genetic examinations existent
- The following previous molecular genetic examinations have been done:

Date / Time of sampling:

Signature:

Specimen requirements and logistics:

- Patient Consent Form for Genetic Testing according to the German Law and Request Form
- Billing information (Insurance-, Institutional- or Selfpay-Billing)
- 5 ml EDTA blood collection tube labeled with patient name and date of birth

Patient name: _____ DOB: _____

- Amyloidosis**
APOA1, B2M, FGA, GSN, LYZ, OSMR, TTR
- Cystic fibrosis**
CFTR
- Fabry disease**
GLA
- Gilbert's syndrome**
MGT1A1
- Hereditary pancreatitis**
CASR, CFTR, CPA1, CTRC, PRSS1, SPINK1
- Hyperlipidemia und Hypercholesterolemia**
ABCA1, ANGPTL3, ANGPTL4, APOA1, APOA5, APOB, APOC2, APOC3, APOE, CETP, EPHX2, GHR, GPIHBP1, LCAT, LDLR, LDLRAP1, LIPC, LIPG, LMF1, LPL, MTP, NPC1L1, PCSK9, PPP1R17
- Hyperlipoproteinemia and Hypercholesterolemia**
ABCA1, ANGPTL3, ANGPTL4, APOA1, APOA5, APOB, APOC2, APOC3, APOE, CETP, EPHX2, GHR, GPIHBP1, LCAT, LDLR, LDLRAP1, LIPC, LIPG, LMF1, LPL, MTP, NPC1L1, PCSK9, PPP1R17
- Hypogonadotropic Hypogonadism, with and without Anosmia**
ANOS1, CHD7, DUSP6, FEZF1, FGF17, FGF8, FGFR1, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, LHB, LHX4, NDNF, NHLH2, NROB1, NR5A1, NSMF, PROK2, PROKR2, PROP1, SEMA3A, SPRY4, TAC3, TACR3, TCF12, WDR11
- Hypogonadotropic Hypogonadism syndromal**
CHD4, CUL4B, DCAF17, DHCR7, GLI2, HFE, LEP, LEPR, POLR3B, RNF216, SEMA3E, SLC29A3, SOX10, SOX2, STS
- Hypoproteinemia**
ABCA1, ANGPTL3, ANGPTL4, APOA1, APOA5, APOB, APOE, CETP, LCAT, LDLR, LIPC, LIPG, LPL, MTP, PCSK9
- Hemochromatosis**
BMP2, FTH1, FTI, HAMP, HFE, HJV, SLC40A1, TFR2
- Inflammatory bowel disease**
ABCB1, ADAM17, ATG16L1, CARD8, DGAT1, EPCAM, FOXP3, GUCY2C, IL10RA, IL10RB, IL23R, IL37, IL6, IRF5, IRGM, INAVA, MVK, MYO5B, NEUROG3, NLRC4, NOD2, PERCC1, PLVAP, SLC26A3, SLC9A3, SPINT2, STX3, ST8SIA2, TGFB1, TTC7A, WNT2B
- Metabolic disorder (whole-Panel)**
(Gene panel with 128 genes)*
- MODY**
ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1
- Mucopolysaccharidosis**
ARSB, GALNS, GBA, GLA, GLB1, GNPTAB, GNPTG, GNS, GUSB, HGSNAT, IDS, IDUA, MCOLN1, NAGLU, NEU1, PSAP, SGSH
- Multiple acyl-CoA dehydrogenase deficiency (DAAD)**
ACADVL, EFTA, ETTB, ETFDH
- Obesity**
ADCY3, ADIPOQ, ADRB2, ADRB3, AGRP, BDNF, CARTPT, CELA2A, CEP19, DYRK1B, ENPP1, FFAR4, FTO, GHRL, GNAS, LEP, LEPR, MC3R, MC4R, MRAP2, NROB2, NTRK2, PCSK1, POMC, PPARG, SDC3, SH2B1, SIM1, UCP1, UCP3
- Periodic fever syndrome**
ELANE, IL1RN, IL36RN, LPIN2, MEFV, MVK, NLRC4, NLRP12, NLRP3, NOD2, PSMB8, PSTPIP1, TMEM173, TNFRSF1A
- Phosphate metabolism disorder**
ALPL, CLCN5, DMP1, ENPP1, FAM20C, FGF23, FGFR1, KL, OCRL, PHEX, SLC34A1, SLC34A3, SLC20A2, SLC9A3R1
- Primary ovarian insufficiency**
AMH, AMHR2, AR, BMP15, BNC1, C14orf39, CLPP, CYP17A1, CYP19A1, DIAPH2, EIF2B2, EIF2B4, EIF2B5, ERAL1, ERCC6, ESR1, FIGLA, FMR1, FOXL2, FSHB, FSHR, GDF9, GGPS1, HARS2, HFM1, HSD17B4, HSF2BP, LARS2, LHB, LHCGR, MCM8, MCM9, MRPS22, MSH5, NOBOX, NR5A1, PANX1, POF1B, PSMC3IP, SOHLH1, SPIDR, STAG3, SYCE1, TWNK, XRCC2, XRCC4, ZP1, ZSWIM7
- Pubertas praecox**
CYP11B1, CYP19A1, GNAS, DLK1, KISS1, KISS1R, LHCGR, MKRN3

* A detailed gene list can be provided on request.