INSTITUTE OF IMMUNOLOGY AND GENETICS

Institute of Immunology and Genetics, Pfaffplatz 10, 67655 Kaiserslautern

LABORATORY MEDICINE
Dr. med. Bernhard Thiele
Prof. Dr. med. Jens Verheyen

HUMAN GENETICS
Prof. Dr. med. Wolfram Henn
Dr. med. Anne-Karin Kahlert

Phone: +49(0)631/31670-0 Fax: +49(0)631/31670-20
office@immungenetik-kl.de | www.immungenetik-kl.de

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, APOE, CETP, GPIHBP1, LCAT, LDLR, LDLRAP1, LIPC, LIPG, LMF1, LPL, MTTP, PCSK9

☐ **Hypoproteinemia**
ABCA1, ANGPTL3, ANGTPL4, APOA1, APOA5, APOB, APOE, CETP, LCAT, LDLR, LIPC, LIPG, LPL, MTTP, PCSK9

☐ **Hemochromatosis**
BMP2, FTH1, FTl, HAMP, HFE, HJV, SLC40A1, TFR2

☐ **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**
ADRB2, ADRB3, AGRP, BDNF, CARTPT, ENPP1, GHRL, LEP, LEPR, MC3R, MC4R, NROB2, NTRK2, PCSK1, POMC, PRARG, SDC3, SIM1, UCP1, UCP3

☐ **Periodc fever syndrome**
ELANE, IL1RN, IL36RN, LPIN2, MEFV, MVK, NLRC4, NLRP12, NLRP3, NOD2, PSMB8, PSTPIP1, TMEM173, TNFRSF1A

☐ **Phosphate metabolism disorer**
ALPL, CLCN5, DMP1, ENPP1, FAM20C, FGF23, FGFR1, KL, OCRL, PHEX, SLC34A1, SLC34A3, SLC20A2, SLC9A3R1

☐ **Primary ovarian insufficiency**
AARS2, BNC1, BMP15, CLPP, CYP17A1, EIF2B5, ERCC6, FANCM, FIGLA, FOXL2, FSHB, FSHR, GALT, GDF9, HFM1, HSD17B4, LARS2, MCM8, MCM9, MRPS22, MSH5, NOBOX, NR5A1, PMM2, SOHLH1, STAG3, TWNK

* A detailed gene list can be provided on request.