Molecular Genetic Testing Request Form

Department of Paediatrics and Adolescent Medicine
Medical Director: Prof. Dr. Miriam Erlacher
Diagnostic Laboratories of the Department of Paediatrics and Adolescent Medicine
University Medical Center Ulm
Molecular Diagnostics Laboratory
Head of Laboratory: Prof. Dr. Ulrich Pannicke
Eythstrasse 24 / D-89075 Ulm / Germany
Telephone: +49 (0)731 500 – 57234 / Fax: +49 (0)731 500 – 57157
e-mail: ulrich.pannicke@uniklinik-ulm.de
https://www.uniklinik-ulm.de/kinder-und-jugendmedizin/labore/diagnostische-laboratorien/molekulardiagnostik-labor.html

DAkkS Deutsche Akkreditierungsstelle D-ML-13294-08-00

Patient data (label if applicable)

_____ _____
Family name, First name(s) Date of birth

_____ _____
Street, Number Postcode

_____ _____
City (Reference/Case number)

Sex: ☐ female ☐ male ☐ diverse

_____
Health insurance fund or responsible payer

Sender

_____ _____
Contact person (capital letters) Telephone

Practice / Clinic stamp with diagnostic report address

_____ _____
Date Signature

Billing information[1]

☐ Statutory health insurance (KV), external outpatient
☐ Privately insured, invoice to patient (please attach billing address)
☐ Internal cost allocation – University Hospital Ulm
☐ Invoice to referring doctor / practice / clinic

[1] If no information is provided, the invoice will be issued to the sender or the patient. The same applies if billing is not possible due to incorrect information.

Sample material*

☐ EDTA blood (3-5 ml)** Date of collection: _____ Time: _____ (2–3 ml if possible for newborns)
☐ DNA sample Volume (µl): _____ Concentration (ng/µl): _____
☐ Other (only with prior agreement): _____
☐ **Second sample (confirmation sample)**

* Exact collection, transport and analysis requirements can be found in our Pre-analytics Manual for the Molecular Diagnostics Laboratory (PDF file). This document is available in German only:
https://www.uniklinik-ulm.de/fileadmin/default/Kliniken/Kinder-Jugendmedizin/Labore/Praeanalytikhandbuch_Molekulare_Diagnostik_Version_7.pdf

** Send without refrigeration

☐ The signed informed consent is enclosed

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Patient information

Indication / Diagnostic Question (with ICD10 code)

ICD10: _____ Indication: _____

☐ Affected individual ☐ Carrier status / Segregation

Stepwise diagnostics requested ☐ No ☐ Yes (please indicate order of steps)

_____

Testing for a known familial pathogenic variant requested

☐ No ☐ Yes (please attach report)

_____ _____

Gene Variant

Family history

☐ Abnormal ☐ Unremarkable ☐ Unknown

(Clinical reports / pedigree sketch requested, if available, on separate sheet)

Genetic findings in the patient: ☐ No ☐ Yes (please attach report) ☐ Unknown

Affected relatives: ☐ No ☐ Yes

Genetic findings in relatives: ☐ No ☐ Yes (please attach report) ☐ Unknown

Consanguinity in the family: ☐ No ☐ Yes

Already examined by us: _____ _____

Name or case number Relationship to the patient

German Genetic Diagnostics Act

According to the German Genetic Diagnostics Act **(GenDG § § 8, 9)**, every genetic test requires counselling of the patient or their legal representative by the responsible physician (requesting party), as well as a written informed consent addressed to the counsellor, including a statement on the retention of unused sample material. Violation of this legal requirement is punishable by law. We can only carry out the requested genetic analysis if the original signed consent form is submitted to us together with this request form.

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	OMIM Gene	Disease / Description	Gene

Immunology / Immune deficiencies

Severe combined immunodeficiency (SCID)

❑	*308380	IL2RG deficiency, X-linked / Interleukin 2 Receptor Subunit Gamma (IL2RG)	*IL2RG*
❑	*147730	IL2RA deficiency, CD25 deficiency / Interleukin 2 Receptor Subunit Alpha (IL2RA)	*IL2RA*
❑	*600173	JAK3 deficiency / Janus Kinase 3 (JAK3)	*JAK3*
❑	*146661	IL7R deficiency / Interleukin 7 Receptor (IL7R)	*IL7R*
❑	*186790	CD3D deficiency / CD3 Delta Subunit Of T-Cell Receptor Complex (CD3D)	*CD3D*
❑	*186830	CD3E deficiency / CD3 Epsilon Subunit Of T-Cell Receptor Complex (CD3E)	*CD3E*
❑	*176947	ZAP70 deficiency, CD8 deficiency / Zeta Chain Of T Cell Receptor Assoc. Protein Kinase 70 (ZAP70)	*ZAP70*

Other defined immunological disorders

❑	*109535	Immunodeficiency with hyper-IgM syndrome, HIGM3 / CD40 Molecule (CD40)	*CD40*
❑	*300386	X-linked hyper-IgM syndrome, HIGM1 / CD40 Ligand (CD40LG)	*CD40LG*
❑	*300392	Wiskott-Aldrich Syndrome, WAS / WASP Actin Nucleation Promoting Factor (WAS)	*WAS*
❑	*170280	Familial haemophagocytic lymphohistiocytosis, FHL / Perforin 1 (PRF1)	*PRF1*
❑	*607358	Autoimmune polyendocrinopathy and ectodermal dysplasia Type 1, APS1, APECED / Autoim. Regul. (AIRE)	*AIRE*
❑	*300292	Immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndr. / Forkhead Box P3 (FOXP3) (IPEX)	*FOXP3*
❑	*107470	Mendelian susceptibility to mycobacterial diseases, MSMD / Interferon Gamma Receptor 1 (IFNGR1)	*IFNGR1*
❑	*134637	Autoimmune lymphoproliferative syndrome, ALPS1a, Canale-Smith syndr. / Fas Cell Surf. Death Recep. (FAS)	*FAS*
❑	*134638	Autoimmune lymphoproliferative syndrome, ALPS1b, Canale-Smith-Syndrom / Fas Ligand (FASLG)	*FASLG*
❑	*601762	Autoimmune lymphoproliferative syndrome, ALPS2, Canale-Smith syndrome / Caspase 10 (CASP10)	*CASP10*
❑	*601763	Autoimmune lymphoproliferative syndrome, ALPS2b, Canale-Smith syndrome / Caspase 8 (CASP8)	*CASP8*
❑	*164790	Autoimmune lymphoproliferative syndrome, ALPS4, N-RAS Exon 2 / NRAS Proto-Oncogene, GTPase (NRAS)	*NRAS*
❑	*300490	X-linked lymphoproliferative syndrome, XLP / SH2 Domain Containing 1A (SH2D1A)	*SH2D1A*
❑	*186973	Autosomal recessive lymphoproliferative disease / IL2 Inducible T Cell Kinase (ITK)	*ITK*
❑	*300079	X-linked lymphoproliferative syndrome / X-Linked Inhibitor of Apoptosis (XIAP) (BIRC4)	*XIAP*
❑	*123890	T-cell function loss / Cytotoxic T-Lymphocyte Associated Protein 4 (CTLA4)	*CTLA4*
❑	*300126	Dyskeratosis congenita type 1, X-linked / Dyskerin Pseudouridine Synthase 1 (DKC1)	*DKC1*
❑	*606609	Aicardi-Goutières syndrome, AGS, CHBL, HERNS, HVR / Three Prime Repair Exonuclease 1 (TREX1)	*TREX1*
❑	*300481	Chronic granulomatous disease, CGD / Cytochrome B-245 Beta Chain (CYBB)	*CYBB*
❑	*608508	Chronic granulomatous disease, CGD / Cytochrome B-245 Alpha Chain (CYBA)	*CYBA*
❑	*608515	Chronic granulomatous disease, CGD / Neutrophil Cytosolic Factor 2 (NCF2)	*NCF2*
❑	*102582	Autosomal dominant hyper-IgE syndrome / Signal Transducer And Activator Of Transcription 3 (STAT3)	*STAT3*
❑	*146933	Inflammatory bowel disease / Interleukin 10 Receptor Subunit Alpha (IL10RA)	*IL10RA*
❑	*123889	Inflammatory bowel disease / Interleukin 10 Receptor Subunit Beta (IL10RB)	*IL10RB*
❑	*124092	Inflammatory bowel disease, Severe infantile colitis / Interleukin 10 (IL10)	*IL10*
❑	*605956	*Blau syndrome, BS / Nucleotide Binding Oligomerization Domain Containing 2 (NOD2)	*NOD2*
❑	*300248	*Incontinentia pigmenti / Inhib. Of Nuclear Factor Kappa B Kinase Regulatory Sub. Gamma (IKBKG) (NEMO)	*IKBKG*

* not accredited

Osteopetrosis

❑	*602727	Osteopetrosis, Malignant infant., Albers-Schönberg disease, ADOII / Chloride Voltage-Gated Chann. 7 (CLCN7)	*CLCN7*
❑	*604592	Osteopetrosis, Malignant infant. / T Cell Immune Regul. 1, ATPase H+ Transp. V0 Sub. A3 (TCIRG1) (OC116)	*TCIRG1*
❑	*607649	Osteopetrosis, Malignant infantile / Osteoclastogenesis Associated Transmembrane Protein 1 (OSTM1)	*OSTM1*
❑	*602642	Osteopetrosis, Osteoclast deficiency / TNF Superfamily Member 11 (TNFSF11) (RANKL)	*TNFSF11*
❑	*603499	Osteopetrosis, Hypogammaglobulinemia / TNF Receptor Superfamily Member 11a (TNFRSF11A) (RANK)	*TNFRSF11A*
❑	*614780	Osteopetrosis, Type B7 / Sorting Nexin 10 (SNX10)	*SNX10*

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	OMIM Gene	Disease / Description	Gene
		Periodic fever syndromes	
❑	*608107	Familial Mediterranean fever, FMF / MEFV Innate Immunity Regulator, Pyrin (MEFV)	*MEFV*
❑	*191190	Familial periodic fever, FPF, TRAPS / TNF Receptor Superfamily Member 1A (TNFRSF1A)	*TNFRSF1A*
❑	*251170	Hyper-IgD syndrome, HIDS / Mevalonate Kinase (MVK)	*MVK*

Haematology

❑	*613609	Hereditary haemochromatosis Type 1 / Homeostatic Iron Regulator (HFE) (HFE1)	*HFE*
❑	*608374	Haemochromatosis Type 2A / Hemojuvelin BMP Co-Receptor (HJV) (HFE2)	*HJV*
❑	*604720	Haemochromatosis Typ3 / Transferrin Receptor 2 (TFR2) (HFE3)	*TFR2*
❑	*604653	Haemochromatosis Typ4 / Solute Carrier Family 40 Member 1 (SLC40A1) (FPN1) (HFE4)	*SLC40A1*
❑	*606464	Haemochromatosis Typ2B, Juvenile Haemochromatosis / Hepcidin Antimicrobial Peptide (HAMP)	*HAMP*
❑	*191740	Meulengracht syndrome, Gilbert syndrome, CNS / UDP Glucuronosyltransferase Family 1 Member A1 (UGT1A1)	*UGT1A1*
❑	*305371	Dyserythropoietic anaemia with thrombocytopenia, X-linked / GATA Binding Protein 1 (GATA1)	*GATA1*
❑	*611184	DHD1, Hereditary xerocytosis / Piezo Type Mechanosensitive Ion Channel Comp. 1 (Er Blood Group) (PIEZO1)	*PIEZO1*
❑	*602754	DHD2, Hereditary xerocytosis / Potassium Calcium-Activated Channel Subfamily N Member 4 (KCNN4)	*KCNN4*
❑	*159530	CAMT, Familial thrombocytosis, Thrombocythemia / MPL Proto-Oncogene, Thrombopoietin Receptor (MPL)	*MPL*
❑	*600044	Familial thrombocytosis, Thrombocythemia / Thrombopoietin (THPO)	*THPO*
❑	*609862	Hereditary therapy-refractory iron deficiency anaemia / Transmembrane Serine Protease 6 (TMPRSS6) (IRIDA)	*TMPRSS6*
❑	*603941	Thiamine-responsive megaloblastic anaemia syndrome / Solute Carrier Family 19 Member 2 (SLC19A2) (TRMA)	*SLC19A2*
❑	*600523	Microcytic anaemia with hepatic iron overload / Solute Carrier Family 11 Member 2 (SLC11A2) (DMT1)	*SLC11A2*
❑	*137295	*GATA2 deficiency spectrum, Acute Myeloid Leukaemia, AML / GATA Binding Protein 2 (GATA2)	*GATA2*
❑	*605577	*Bleeding disorder due to CalDAG-GEFI deficiency / RAS Guanyl Releasing Protein 2 (RASGRP2)	*RASGRP2*

* not accredited

Endocrinology and Diabetology

		Disorders of glucose regulation	
❑	*600281	Maturity-Onset Diabetes of the Young, MODY Type 1 / Hepatocyte Nuclear Factor 4 Alpha (HNF4A) (MODY1)	*HNF4A*
❑	*138079	Maturity-Onset Diabetes of the Young, MODY Type 2 / Glucokinase (GCK) (MODY2)	*GCK*
❑	*142410	Maturity-Onset Diabetes of the Young, MODY Type 3 / HNF1 Homeobox A (HNF1A) (TCF1) (MODY3)	*HNF1A*

Molecular genetic analyses of genes not listed here can also be performed upon request.

For any questions, please consult the designated specialist contact in all cases.

Contact person (☎ Telephone, e-mail)

Immunology: Prof. Dr. Ansgar Schulz, ☎ +49 (0)731 500-57174, ansgar.schulz@uniklinik-ulm.de

Haematology: Prof. Dr. H. Cario, ☎ +49 (0)731 500-57219, holger.cario@uniklinik-ulm.de

Endocrinology and Diabetology: Prof. Dr. M. Wabitsch, ☎ +49 (0)731 500-57400, martin.wabitsch@uniklinik-ulm.de