

Diagnostic Laboratories Department of Pediatrics and Adolescent Medicine

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Informed consent of patient

The German law (**GenDiagnostikgesetz – GenDG**) defines under which circumstances genetic testing of a human individual is legal. *Diagnostic* testing can only be conducted with the patient's informed consent which requires documented consultation with a doctor. *Predictive* testing requires genetic counselling by a human genetics specialist prior to and after the investigation, or the patient's written renunciation.
Please fill out the informed consent carefully and completely and sign it. Otherwise no molecular genetic testing can be performed!

Molecular Genetic Testing

3-5 ml EDTA-blood (send by normal mail)

Turnaround time: 2-4 weeks, more than one analysis and rare analyses may take up to 8 weeks



OMIM	Disease	Gene
Immunology		
Immunodeficiency SCID (T-B+)		
<input type="checkbox"/>	OMIM 300400 Severe Combined Immunodeficiency (SCID, X-chromosomal, T-, B+, NK-)	IL2RG
<input type="checkbox"/>	OMIM 606367 Severe Combined Immunodeficiency, CD25 deficiency	IL2RA
<input type="checkbox"/>	OMIM 600802 Severe Combined Immunodeficiency (SCID, T-, B+, NK-), JAK3 deficiency	JAK3
<input type="checkbox"/>	OMIM 608971 Severe Combined Immunodeficiency (SCID, T- B+ NK+), IL7R deficiency	IL7R
<input type="checkbox"/>	OMIM 608971 Severe Combined Immunodeficiency (SCID, T- B+ NK+), CD3D deficiency	CD3D
<input type="checkbox"/>	OMIM 608971 Severe Combined Immunodeficiency (SCID, T- B+ NK+), CD3E deficiency	CD3E
Other defined immune disorders		
<input type="checkbox"/>	OMIM 109535 Immunodeficiency - Hyper-IgM-Syndrom (HIGM 3)	CD40
<input type="checkbox"/>	OMIM 300386 Hyper-IgM-Syndrom, X-linked (HIGM 1), (SCID, T- B-)	CD40L
<input type="checkbox"/>	OMIM 176947 Severe Combined Immunodeficiency (SCID, T-, B-), ZAP70 deficiency, CD8 deficiency	ZAP70
<input type="checkbox"/>	OMIM 301000 Wiskott-Aldrich Syndrome	WASP
<input type="checkbox"/>	OMIM 603553 familial hemophagocytic lymphohistiocytosis (FHL2; HLH2)	PRF1
<input type="checkbox"/>	OMIM 240300 autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APS1), APECED	AIRE
<input type="checkbox"/>	OMIM 304790 immune dysregulation, polyendocrinopathy, enteropathy, X-linked (IPEX)	FOXP3
<input type="checkbox"/>	OMIM 209950 familial disseminated atypical mycobacterial infection and disseminated BCG infection (FDAMI)	IFNGR1
<input type="checkbox"/>	OMIM 601859 Autoimmune lymphoproliferative syndrome (ALPS1a), Canale-Smith-Syndrom	AP01
<input type="checkbox"/>	OMIM 601859 Autoimmune lymphoproliferative syndrome (ALPS1b), Canale-Smith-Syndrom	FASL
<input type="checkbox"/>	OMIM 603909 Autoimmune lymphoproliferative syndrome (ALPS2a), Canale-Smith-Syndrom	CASP10
<input type="checkbox"/>	OMIM 607271 Autoimmune lymphoproliferative syndrome (ALPS2b), Canale-Smith-Syndrom	CASP8
<input type="checkbox"/>	OMIM 164790 Autoimmune lymphoproliferative syndrome(ALPS4); N-RAS exon 2	N-Ras
<input type="checkbox"/>	OMIM 308240 X-linked lymphoproliferative syndrome (XLP), Purtillo-Syndrom (SH2D1A)	SAP
<input type="checkbox"/>	OMIM 300248 incontinentia pigmenti, hypohidrotic ectodermal dysplasia, (EDA-ID); X-chrom. recessive; <i>not: Incontinentia Pigmenti -del. Ex 4-10</i>	NEMO
<input type="checkbox"/>	OMIM 613011 EBV-associated lymphoproliferative diseases, autosomal-rezessive; IL2-inducible T-cell kinase, ITK deficiency	ITK
<input type="checkbox"/>	OMIM 300079 X-linked lymphoproliferative syndrome (XLP2), BIRC4 deficiency	BIRC4
<input type="checkbox"/>	OMIM 154545 infections, recurrent, with chronic diarrhea and opsonisation defect (low MBL2), including susceptibility to	MBL2
<input type="checkbox"/>	OMIM 123890 Graves disease, susceptibility locus 5 (GD5S), T-cell defect	CTLA4
<input type="checkbox"/>	OMIM 305000 Dyskeratosis congenita; X-linked; Codon: p.A353V	DKC1
<input type="checkbox"/>	OMIM 305000 Dyskeratosis congenita-1; X-linked	DKC1
<input type="checkbox"/>	OMIM 266100 epilepsy, pyridoxine-dependent (EPD); pyridoxine dependency with seizures, ALDH7A1 deficiency	ALDH7A1
<input type="checkbox"/>	OMIM 606609 Aicardi-Goutieres Syndroms AGS, chilblain lupus CHBL, hereditary endotheliopathy with retinopathy, nephropathy	TREX1
<input type="checkbox"/>	OMIM 300645 chronic granulomatous disease 1 (CGD1); X-linked granulomatous disease; cytochrome b-245, beta polypeptide	CYBB
<input type="checkbox"/>	OMIM 608508 CGD; Chronic Granulomatose; Cytochrome b-245 Alpha Kette - CYBA	CYBA
<input type="checkbox"/>	OMIM 612301 CGD; Chronic Granulomatose; neutrophiler cytosolischer Factor 2 - NCF2	NCF2
<input type="checkbox"/>	OMIM 102582 Hyper-IgE-Syndrom, autosomal-dominantes (Exon 13, 21 und 23; p.K392R; p.N646K; p.K658N; p.T715M)	STAT3
Osteopetrosis		
<input type="checkbox"/>	OMIM 602727 osteopetrosis, Albers-Schonberg disease 4 (OPTB4), recessive	CLCN7
<input type="checkbox"/>	OMIM 602727 osteopetrosis, type II (OPTA2), autosomal dominant, ADOII, Albers-Schoenberg disease	CLCN7
<input type="checkbox"/>	OMIM 604592 osteopetrosis, lethal B1 (OPTB1)	TCIRG1
<input type="checkbox"/>	OMIM 607649 osteopetrosis type 1B 5 (OPTB5), malignant infantile	OSTM1
<input type="checkbox"/>	OMIM 602642 osteopetrosis type B2 (POTB2), TNFSF11 deficiency	RANKL
<input type="checkbox"/>	OMIM 612301 Osteopetrose, (TNFRSF11A), Hypogammaglobulinämie	RANK
<input type="checkbox"/>	OMIM 612301 Osteopetrose, (SNX10), Typ B7	SNX10
Periodic Fever Syndromes		
<input type="checkbox"/>	OMIM 249100 Familial Mediterranean fever (FMF)	MEFV
<input type="checkbox"/>	OMIM 142680 TNF receptor associated periodic syndrome, benign autosomal dominant familial periodic fever (FPF), Hibernian fever	TRAPS
<input type="checkbox"/>	OMIM 260920 Hyperimmunoglobulinemia D with recurrent fever (HIDS), mevalonate kinase deficiency	MVK
Hematology		
<input type="checkbox"/>	OMIM 300367 dyserythropoietic anemia with thrombocytopenia (X-linked), CDATX	GATA-1
<input type="checkbox"/>	OMIM 603474 Diamond-Blackfan anemia (DBA)	RPS19
<input type="checkbox"/>	OMIM 603634 Diamond-Blackfan anemia (DBA5)	RPL5
<input type="checkbox"/>	OMIM 611184 Dehydrated hereditary Stomatocytosis 1 (DHD1); Hereditary Xerozytosis	PIEZO1
<input type="checkbox"/>	OMIM 602754 Dehydrated hereditäre Stomatocytosis 2 (DHD2); Hereditary Xerozytosis	KCNN4
<input type="checkbox"/>	OMIM 159530 congenital amegakaryocytic thrombocytopenia (CAMT)	MPL
<input type="checkbox"/>	OMIM 600044 thrombocythemia, essential (THPO)	THPO

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Hematology		
<input type="checkbox"/>	OMIM 206200 iron-refractory iron deficiency anemia (IRIDA)	TMPRSS6
<input type="checkbox"/>	OMIM 600424 disturbance in MTX clearance, high MTX levels	SLC19A1/GGH
<input type="checkbox"/>	OMIM 249270 thiamine responsive megaloblastic anemia (TRMA)	SLC19A2
<input type="checkbox"/>	OMIM 600523 pseudo-iron deficiency anemia (PIDA), microcytic anemia with liver iron overload, DMT1 deficiency	SLC11A2/DMT1
<input type="checkbox"/>	OMIM147700 isocitrate dehydrogenase 1 (NADP+), soluble, associated with brain tumors and secondary Glioblastoma	IDH1
<input type="checkbox"/>	OMIM 613657 isocitrate dehydrogenase 2 (NADP+), mitochondrial; D-2-hydroxyglutaric aciduria (D2HA2)	IDH2
<input type="checkbox"/>	OMIM 613673 congenital dyserythropoietic anemia, TYPE IV; CDAN4	KLF1

Endocrinology and Diabetology

Disturbance in glucose regulation

<input type="checkbox"/>	OMIM 125850 Maturity-Onset Diabetes of the Young, MODY-Diabetes, MODY type 1 (MODY1)	HNPF4a
<input type="checkbox"/>	OMIM 125851 Maturity-Onset Diabetes of the Young, MODY-Diabetes, MODY type 2 (MODY2)	GCK
<input type="checkbox"/>	OMIM 600496 Maturity-Onset Diabetes of the Young, MODY-Diabetes, MODY type 3 (MODY3)	HNPF1a/TCF1

Disturbance in body weight regulation

<input type="checkbox"/>	OMIM 155541 severe obesity (OBS3)	MC4R
<input type="checkbox"/>	OMIM 164160 severe early-onset obesity (OBS1)	LEP
<input type="checkbox"/>	OMIM 601007 early-onset morbid obesity (only after having consulted Prof. Wabitsch)	LEPR
<input type="checkbox"/>	OMIM 601047 Berardinelli-Seip congenital generalized lipodystrophy type (BSCL3)	CAV1
<input type="checkbox"/>	OMIM 613327 congenital generalized lipodystrophy type 4 (CGL4)	PTRF

Disturbance of growth

<input type="checkbox"/>	OMIM 117550 Sotos syndrome (STO)	NSD1
<input type="checkbox"/>	OMIM 610978 congenital thyroglobulin defect with respiratory distress (TGDRD)	NKX2-1
<input type="checkbox"/>	OMIM 118700 benign hereditary chorea (BHC)	NKX2-1

Polymorphisms (possibly clinical relevant)

<input type="checkbox"/>	OMIM 174800 McCune-Albright Syndrome (MAS)	GNAS
<input type="checkbox"/>	OMIM 167413 Diabetes	PAX4
<input type="checkbox"/>	OMIM 173360 Obesity	PAI1
<input type="checkbox"/>	OMIM 601487 Obesity and thromboembolism	PPARG2
<input type="checkbox"/>	OMIM 151750 Obesity	HSL
<input type="checkbox"/>	OMIM 170290 Obesity	PLIN
<input type="checkbox"/>	OMIM 190220 Obesity and Diabetes	TGFb1
<input type="checkbox"/>	OMIM 601283 Obesity; Diabetes Mellitus, Noninsulin-dependent, 1 (NIDDM1)	CAPN10
<input type="checkbox"/>	OMIM 605441 Obesity, Adipocyte, C1q, and Collagen Domain containing,(ADIPOQ)	APM1
<input type="checkbox"/>	OMIM 601693 Obesity	UCP-2
<input type="checkbox"/>	OMIM 107720 Insulin resistance in nonalcoholic fatty liver disease (c.-482C>T; c.-455T>C)	APOC3
<input type="checkbox"/>	OMIM 600716 Diabetes (type 1)	PtPN22

Gastroenterology

<input type="checkbox"/>	OMIM 608374 hemochromatosis, type 2, Hämojuvelin (HVJ)	HFE2
<input type="checkbox"/>	OMIM 604250 hemochromatosis, type 3; transferrin receptor 2, TFR2	HFE3/TFR2
<input type="checkbox"/>	OMIM 606069 hemochromatosis, type 4; solute carrier family 40 (iron-regulated transporter), member 1, FPN1 / SLC40A1	HFE4/FPN1
<input type="checkbox"/>	OMIM 602390 hemochromatosis, juvenile, (JH), (HFE2B)	HAMP
<input type="checkbox"/>	OMIM 143500 Gilbert-Syndrom (GBTS)	UGT1A1
<input type="checkbox"/>	OMIM 146933 chronic inflammatory bowel disease, interleukin 10 receptor, alpha; IL10RA	IL10Ra
<input type="checkbox"/>	OMIM 123889 chronic inflammatory bowel disease, interleukin 10 receptor, beta; IL10RB	IL10Rb
<input type="checkbox"/>	OMIM 124092 chronic inflammatory bowel disease, interleukin 10; early onset	IL10
<input type="checkbox"/>	OMIM 610370 congenital malabsorptive diarrhea 4 (DIAR4); T2DM; hyperproinsulinaemia	NEUROG3

Polymorphisms (possibly clinical relevant)

<input type="checkbox"/>	OMIM 266600 chronic inflammatory bowel disease, 1 (IBD1), Morbus Crohn	NOD2
<input type="checkbox"/>	OMIM 605956 Hepatic veno-occlusive disease or veno-occlusive disease (VOD)	NOD2
<input type="checkbox"/>	OMIM 235200 hemochromatosis (HFE1); polymorphism p.H63D, p.S65C und p.C282Y	HFE

Information about the indication of molecular diagnosis can be achieved via **“The portal for rare diseases and orphan drugs“**
-orphanet- : <http://www.orpha.net/consor/cgi-bin/index.php> or http://www.orpha.net/consor/cgi-bin/Disease_Genes.php?lng=EN

External quality assurance (EQA):

our laboratory participates regularly on „Ringversuche“ of the national Reference Institute for Bioanalytics (RfB). These RfB-laboratories determine method-independent target values, so called reference methods values, in control materials for external quality control.