

## Diagnostik Algorithmen - medgen.at GmbH

Angebot Diagnostik Molekulargenetik (**M**)/ Zytogenetik (**Z**)/ FISH (**F**)

Erkrankung	OMIM	Genlocus	OMIM	M	Z	F
Achondroplasie	100800	<i>FGFR3</i>	134934	x		
ADAM17		<i>ADAM17</i>	603639	x		
Adrenogenitales Syndrom bei 21-Hydroxylasemangel (Schnelltest und Komplettanalyse)	201910	<i>CYP21A2</i>	613815	x	(x)	
Alagille Syndrom 1	118450	<i>JAG1</i>	601920	x		
Amyotrophe Lateralsklerose 2	205100	<i>ALS2</i>	606352	x		
Angelman Syndrom	105830	<i>SNRPN</i>	182279	x	x	x
APECED	240300	<i>AIRE</i>	607358	x		
Apert Syndrom	101200	<i>FGFR2</i>	176943	x		
Autoimmunthyroiditis		<i>CTLA4</i> c.49G>A (rs231775)	123890	x		
Azoospermie, Oligospermie	415000	<i>AZF</i>	415000	x	x	
Beckwith-Wiedemann Syndrom	130650	<i>H19/KCNQ1OT1</i>	103280/604115	x	x	
Bernard-Soulier Syndrom	231200	<i>GP1BA/GP1BB/GP9</i>	606672/138720/173515	x		
Blackfan-Diamond Anämie	105650	<i>RPS19</i>	603474	x		
Blackfan-Diamond Anämie Whole Exome Sequencing (WES, 21 Gene) und MLPA	105650	<i>GATA1/RPL11/RPL15/RPL26/RPL27/RPL31/RPL35A/RPL36/RPL5/RPS10/RPS15/RPS17/RPS19/RPS24/RPS26/RPS27/RPS27A/RPS28/RPS29/RPS7/TSR2</i>	305371/604175/604174/603704/607526/-/180468/-/603634/603632/180535/180472/603474/602412/603701/603702/191343/603685/603633/603658/300945	x		
BOR Syndrom Whole Exome Sequencing (WES, 3 Gene)	113650	<i>EYA1/SIX5/SIX1</i>	601653/600963/601205	x		
Börjeson-Forssman-Lehmann Syndrom	301900	<i>PHF6</i>	300414	x		
CACP Syndrom	208250	<i>PRG4</i>	604283	x		
Carney Complex	160980	<i>PRKAR1A</i>	188830	x		
Charcot-Marie-Tooth Erkrankung Typ 2A	609260	<i>MFN2</i>	608507	x		
Charcot-Marie-Tooth Erkrankung Typ 4C	601596	<i>SH3TC2</i>	608206	x		
Charcot-Marie-Tooth Erkrankung Typ 4H	609311	<i>FGD4</i>	611104	x		
Charge Syndrom 1 Whole Exome Sequencing (WES)	214800	<i>CHD7</i>	608892	x	x	
Congenitale amegakaryozytische Thrombopenie (CAMT)	604498	<i>MPL</i>	159530	x		
Congenitales nephrotisches Syndrom	256300/ 600995/ 256370	<i>NPHS1/NPHS2/WT1</i>	602716/604766/607102	x		

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Cowden Syndrom	158350	<i>PTEN</i>	601728	x		
Crigler-Najjar Syndrom (Typ I, II)	218800/ 606785	<i>UGT1A1</i>	191740	x		
Crouzon Syndrom	123500	<i>FGFR2</i>	176943	x		
Crouzon Syndrom <b>mit Akantosis nigricans</b>	612247	<i>FGFR3</i>	134934	x		
CSF3R Exons 14, 17		<i>CSF3R</i>	138971	x		
CVID (TNFRSF13B)	240500	<i>TNFRSF13B</i>	604907	x		
CVID (TNFRSF13C)	240500	<i>TNFRSF13C</i>	606269	x		
Cystische Fibrose	219700	<i>CFTR</i>	602421	x		
Denys-Drash Syndrom	194080	<i>WT1</i>	607102	x		
Distale renale tubuläre Azidose (DRTA), SLC4A1-assoziiert	179800/ 611590	<i>SLC4A1</i>	109270	x		
Ektodermale Dysplasie (WES, 42 Gene)				x		
Ektodermale Dysplasiesyndrome, TP63-assoziiert	604292	<i>TP63</i>	603273	x		
Epilepsie Whole Exome Sequencing (WES, ca. 340 Gene)				x		
Erythropoietische Protoporphyrrie	177000	<i>FECH</i>	612386	x		
Faktor V Leiden	227400	<i>F5</i>	612309	x		
Familiäre adenomatöse Polyposis coli 1 und 2 Whole Exome Sequencing (WES)	175100/ 608456	<i>APC/MUTYH</i>	611731/604933	x		
Familiäre dysalbuminämische Hyperthyroxinämie	615999	<i>ALB</i>	103600	x		
Familiäre Erythrocytose 1	133100	<i>EPOR</i>	133171	x		
Familiäre Granulomatosesyndrome (Blau Syndrom, Early-onset Sarcoidosis)	186580/ 609464	<i>NOD2</i>	605956	x		
Familiäre hypocalciurische Hypercalcämie Typ 1	145980	<i>CASR</i>	601199	x		
Familiäre hypocalciurische Hypercalcämie Typ 2	145981	<i>GNA11</i>	139313	x		
Familiäre hypocalciurische Hypercalcämie Typ 3	600740	<i>AP2S1</i>	602242	x		
Familiärer isolierter Hyperparathyreoidismus	145000	<i>CDC73 (=HRPT2)</i>	607393	x		
Familiäres Mittelmeerfieber	249100	<i>MEFV</i>	608107	x		
Fragiles X Syndrom	300624	<i>FMR1</i>	309550	x	x	
Fragiles X Tremor/Ataxie Syndrom	300623	<i>FMR1</i>	309550	x		
Frasier Syndrom	136680	<i>WT1</i>	607102	x		
GATA1 assoziierte Zytopenie		<i>GATA1</i>	305371	x		

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GATA2		<i>GATA2</i>	137295	x		
Glucose-6-Phosphat-Dehydrogenasemangel		<i>G6PD</i>	305900	x		
Gonadendysgenese (LHCGR)		<i>LHCGR</i>	152790	x		
Gonadendysgenese (NR5A1 (=SF1))		<i>NR5A1 (=SF1)</i>	184757	x		
Gonadendysgenese (SRY)		<i>SRY</i>	480000	x	x	x
Hämochromatose	235200	<i>HFE</i>	613609	x		
Hämoglobinopathien	604131/ 613985/ 603903	<i>HBB/HBA1/HBA2</i>	141900/141800/141850	x		
Hereditäre Albright'sche Osteodystrophie (AHO)/Pseudohypoparathyreoidismus	103580	<i>GNAS</i>	139320	x		
Hereditäre Pankreatitis	167800	<i>SPINK1/PRSS1/CFTR/CTRC</i>	167790/276000/ 602421/601405	x		
Holt-Oram Syndrom	142900	<i>TBX5</i>	601620	x		
Hyper-IgD Syndrom	260920	<i>MVK</i>	251170	x		
Hyper-IgE Syndrom, dominant	147060	<i>STAT3</i>	102582	x		
Hyper-IgE Syndrom, rezessiv	243700	<i>DOCK8</i>	611432	x		
Hyper-IgM Immundefizienz, X-chromosomal	308230	<i>CD40LG</i>	300386	x		
Hyperbilirubinämie Typ 1	237900	<i>UGT1A1</i>	191740	x		
Hyperhomocysteinämie	603174	<i>MTHFR</i>	607093	x		
Hypocalcämie autosomal dominant/ Hypoparathyreoidismus	615361	<i>GNA11</i>	139313	x		
Hypochondroplasie	146000	<i>FGFR3</i>	134934	x		
Hypothyreose, Schilddrüsendyshormonogenese	274500	<i>TPO</i>	606765	x		
IL7R Immundefizienz	608971	<i>IL7R</i>	146661			
IPEX Syndrom	304790	<i>FOXP3</i>	300292	x		
Kallmann Syndrom Whole Exome Sequencing (WES, 39 Gene)		u.a. <i>KAL1/FGFR1/PROKR2/ PROK2/CHD7/FGF8</i>	300836/136350/607123/ 607002/608892/600483	x		
Laktoseintoleranz, primäre Form	223100	<i>LCT</i> rs4988235 (-13910T>C), rs182549 (-22018A>G)	601806	x		
LIG4 Syndrom	606593	<i>LIG4</i>	601837	x		
Lissenzephalie Whole Exome Sequencing (WES, mind. 15 Gene) und MLPA		u.a. <i>ADGRG1(=GPR56)/ARX/DCX/FKRP/ FKTN/LARGE/LIS1/PAFAH1B1/ POMGnT1/POMT1/POMT2/RELN/ TUBA1A/TUBB2B/VLDLR</i>	604110/300382/300121/606596/ 607440/-/-/601545/ 606822/607423/607439/600514/ 602529/612850/192977	x		
Lissenzephalie Typ 1, autosomal	607432	<i>PAFAH1B1</i>	601545	x		

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Lissenzephalie Typ 1, X-chromosomal (Double Cortex Syndrom)	300067	<i>DCX</i>	300121	x		
Lissenzephalie Typ 3, autosomal	611603	<i>TUBA1A</i>	602529	x		
Marfan Syndrom Whole Exome Sequencing (WES, 4 Gene)		<i>FBN1/TGFB2/TGFBR1/TGFBR2</i>	134797/190220/190181/190182	x		
MBL2 Defizienz	614372	<i>MBL2</i>	154545	x		
Methämoglobinämie (Typ I, II)	250800	<i>CYB5R3</i>	613213	x		
Mikrodeletions-, Mikroduplikationssyndrome				x	x	
Maturity-onset diabetes of the young (MODY) Typ 2	125851	<i>GCK</i>	138079	x		
Maturity-onset diabetes of the young (MODY) Typ 3	600496	<i>HNF1A</i>	142410	x		
Muckle Wells Syndrom	191900	<i>NLRP3</i>	606416	x		
Multiple endokrine Neoplasie Typ 1 (MEN1)	131100	<i>MEN1</i>	613733	x		
Multiple endokrine Neoplasie Typ 1 (MEN1)-assoziierte Erkrankungen (WES, 10 Gene) und MLPA		<i>AIP/AP2S1/CASR/CDC73/CDKN1A/CDKN1B/CDKN2B/CDKN2C/GNA11/MEN1</i>	605555/602242/601199/607393/116899/600778/600431/603369/139313/613733	x		
Multiple endokrine Neoplasie Typ 2A (MEN2A)	171400	<i>RET</i> -Protoonkogen	164761	x		
Multiple endokrine Neoplasie Typ 2B (MEN2B)	162300	<i>RET</i> -Protoonkogen	164761	x		
Multiple endokrine Neoplasie Typ 4 (MEN4)	610755	<i>CDKN1B</i>	600778	x		
Multiple kartilaginäre Exostosen	133700	<i>EXT1/EXT2</i>	608177/608210	x		
Myeloproliferative Erkrankungen mit Erythrozytose		<i>JAK2</i>	147796	x	x	
MYH9 assoziierte Makrothrombozytopenien (head bzw tail domain Mut; inkl MLPA)		<i>MYH9</i>	160775	x		
MYH9 assoziierte Makrothrombozytopenien Whole Exome Sequencing (WES) und MLPA		<i>MYH9</i>	160775	x		
Nijmegen-Chromosomalbruch Syndrom	251260	<i>NBS1</i>	602667	x		
Noonan Syndrom Whole Exome Sequencing (WES, 19 Gene)	163950	<i>PTPN11/SOS1/KRAS/RAF1/NRAS/BRAF/RIT1/MAP2K1/SOS2/SHOC2/CBL/LZTR1/SPRED1/HRAS/MAP2K2/NF1/RASA2/RRAS/A2ML1</i>	176876/182530/190070/164760/164790/164757/609591/176872/601247/602775/165360/600574/609291/190020/601263/613113/601589/165090/610627	x	x	
NUDT15 assoziierte Leukopenie	616903	<i>NUDT15</i>	615792	x		
Osteogenesis imperfecta Whole Exome Sequencing (WES, 14 Gene)		<i>COL1A1/COL1A2/BMP1/CRTAP/FKBP10/IFITM5/P3H1/PP1B/SERPINF1/SERPINH1/SP7/SPARC/TMEM38B/WNT1</i>	120150/120160/112264/605497/607063/614757/610339/123841/172860/600943/606633/182120/611236/164820	x		
Osteopetrosis, autosomal rezessiv bzw. dominant	259700/ 611490/ 166600/ 259720	<i>TCIRG1/CICN7/OSTM1</i>	604592/602727/607649	x		
Pachydermoperiostose	259100	<i>HPGD</i>	601688	x		
Paragangliom 1 (PGL1)	168000	<i>SDHD</i>	602690	x		

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Paragangliom 3 (PGL3)	605373	<i>SDHC</i>	602413	x		
Paragangliom 4 (PGL4)	115310	<i>SDHB</i>	185470	x		
Paroxysmale Dyskinesie Whole Exome Sequencing (WES, 10 Gene)		<i>ADCY5/CHRNA2/CHRNA4/CHRN2/ KCNA1/KCNT1/PNKD/PRRT2/ SCN8A/SLC2A1</i>	600293/118502/118504/118507/ 600150/608167/609023/614386/ 600702/138140	x		
Pelizaeus-Merzbacher Erkrankung	312080	<i>PLP1</i>	300401	x	x	
Pena Shokeir Syndrom Typ I/congenitales myasthenes Syndrom	208150	<i>RAPSN</i>	601592	x		
Pendred Syndrom	274600	<i>SLC26A4</i>	605646	x		
Periodisches Fieber, autosomal dominant (TRAPS)	142680	<i>TNFRSF1A</i>	191190	x		
Pfeiffer Syndrom	101600	<i>FGFR2/FGFR1</i>	176943/136350	x		
Phäochromozytom/Paragangliom Whole Exome Sequencing (WES, mind. 20 Gene) und MLPA	171300	u.a. <i>RET/SDHD/SDHB/SDHC/VHL/ SDHA/SDHAF2/MAX/TMEM127/ IDH1/NF1/KIF1B/EGLN1/EPAS1/FH/ EGLN2/GDNF/IDH2/PRKAR1A/HRAS</i>	164761/602690/185470/602413/608537/ 600857/613019/154950/613403/147700/ 613113/605995/606425/603349/136850/ 606424/600837/147650/188830/190020	x		
Prader Willi Syndrom	176270	<i>SNRPN</i>	182279	x	x	x
Premature Ovarian Failure 1 (FMR1)	311360	<i>FMR1</i>	309550	x		
Premature Ovarian Failure 4 (BMP15)	300510	<i>BMP15</i>	300247	x		
Progressive myoklonale Epilepsie des Typs Unverricht Lundborg	254800	<i>CSTB</i>	601145	x		
Prothrombin		<i>F2</i>	176930	x		
Pseudoachondroplasie	177170	<i>COMP</i>	600310	x		
Pseudohypoadosteronismus Typ I	177735	<i>NR3C2 (=MLR)</i>	600983	x		
Pyruvatkinasemangel	266200	<i>PKLR</i>	609712	x		
RAG1 Immundefizienz		<i>RAG1</i>	179615	x		
RAG2 Immundefizienz		<i>RAG2</i>	179616	x		
Rett (like) Syndrom Whole Exome Sequencing (WES, 5 Gene) und MLPA		<i>MECP2/CDKL5/FOXP1/MEF2C/NTNG1</i>	300005/300203/164874/600662/608818	x		
Rett Syndrom, MECP2 Duplikationssyndrom	312750/ 300260	<i>MECP2</i>	300005	x	x	
Robinow Syndrom, autosomal rezessiv	268310	<i>ROR2</i>	602337	x		
Rubinstein Taybi Syndrom Typ 1	180849	<i>CREBBP</i>	600140	x		
RUNX1	601399	<i>RUNX1</i>	151385	x		
Schilddrüsenhormonresistenz	188570	<i>THRB</i>	190160	x		
SETBP1 Mutationshotspots im Exon 4		<i>SETBP1</i>	611060	x		
SH2B3-assoziierte Erythrozytose	133100	<i>SH2B3</i>	605093	x		
SHOX-assoziiertes Haploinsuffizienzsyndrom		<i>SHOX</i>	312865	x	x	x

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SIFD Syndrom	616084	<i>TRNT1</i>	612907	x		
Silver-Russell Syndrom	180860	<i>H19/KCNQ1OT1/PEG1 (=MEST) (UPD 7)</i>	103280/604115/601029	x	x	
Simpson-Golabi-Behmel Syndrom 1	312870	<i>GPC3</i>	300037	x		
Smith-Lemli-Opitz Syndrom/Autismus	270400	<i>DHCR7</i>	602858	x		
Sotos Syndrom	117550	<i>NSD1</i>	606681	x	x	
Subtelomeranalyse				x	x	
Syndrom des persistenten Müller-Ganges Typ II	261550	<i>AMHR2</i>	600956	x		
Taubheitsassoziierte distale renale tubuläre Azidose (DRTA)	267300	<i>ATP6V1B1</i>	192132	x		
Thiamin-responsive megaloblastische Anämie (TRMA)	249270	<i>SLC19A2</i>	603941	x		
Thrombocytose (CALR Exon 9)	187950	<i>CALR</i>	109091	x		
Thrombocytose (MPL Exon 2-4, 10-11)		<i>MPL</i>	159530	x		
Torsionsdystonie, autosomal dominant	128100	<i>TOR1A (=DYT1)</i>	605204	x		
Transthyretin Amyloidose	176300	<i>TTR</i>	176300	x		
Tumoröse Calcinose, familiär hyperphosphatämisch	211900	<i>FGF23/GALNT3/KL</i>	605380/601756/604824	x		
Tumoröse Calcinose, familiär normophosphatämisch	610455	<i>SAMD9</i>	610456	x		
Uniparentale Disomie 14		<i>MEG3</i>	605636	x	x	
Vitamin D-abhängige Rachitis Typ I (Vitamin D-1 alpha-Hydroxylasedefizienz)	264700	<i>CYP27B1</i>	609506	x		
Von Hippel Lindau Syndrom	193300	<i>VHL</i>	608537	x		
Wachstumshormoninsensitivitätssyndrom	262500	<i>GHR</i>	600946	x		
Wachstumsstörung (GH1)		<i>GH1</i>	139250	x		
Wachstumsstörung (GHRH)		<i>GHRH</i>	139190	x		
Wachstumsstörung (GHRHR)		<i>GHRHR</i>	139191	x		
Wachstumsstörung (HESX1)		<i>HESX1</i>	601802	x		
Wachstumsstörung (IGF1)		<i>IGF1</i>	147440	x		
Wachstumsstörung (IGF1R)		<i>IGF1R</i>	147370	x		
Wachstumsstörung (IGFALS)		<i>IGFALS</i>	601489	x		
Wachstumsstörung (POU1F1)		<i>POU1F1 (=PIT1)</i>	173110	x		
Wachstumsstörung (PROP1)		<i>PROP1</i>	601538	x		
Wachstumsstörung (WES, 12 Gene) und MLPA		<i>GH1/GHR/GHRH/GHRHR/HESX1/IGF1/IGF1R/IGFALS/LHX3/LHX4/POU1F1(=PIT1)/PROP1</i>	139250/600946/139190/139191/601802/147440/147370/601489/600577/602146/173110/601538	x		

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WHIM Syndrom	193670	<i>CXCR4</i>	162643	x		
Whole Exome Sequencing (WES)				x		
Wilms Tumor 1	194070	<i>WT1</i>	607102	x		
Wiskott Aldrich Syndrom	301000	<i>WAS</i>	300392	x		
X-gebundene Agammaglobulinämie	300755	<i>BTK</i>	300300	x		
X-gebundene lymphoproliferative Erkrankung Typ 1 (XLP 1)	308240	<i>SH2D1A</i>	300490	x	x	
X-gebundene lymphoproliferative Erkrankung Typ 2 (XLP 2)	300635	<i>XIAP</i>	300079	x		
X-gebundene mentale Retardierung		<i>AGTR2 ARHGEF6GDI1 ARX DCX FACL4  FMR1 FMR2 IL1RAPL1 OPHN1 PAK3  PQBP1 RPS6KA3 SLC6A8 TM4SF2</i>		x	x	
X-gebundener renaler Diabetes insipidus	304800	<i>AVPR2</i>	300538	x		
X Inaktivierung		<i>AR/FMR1</i>	313700/309550	x		
X-SCID	300400	<i>IL2RG</i>	308380	x		
ZAP70 Defizienz	269840	<i>ZAP70</i>	176947	x		
Zöliakie (HLA DQ2, DQ8)	212750	<i>HLA (DQ2, DQ8)</i>	146880/604305	x		
Zuckerintoleranz (Laktose-, Fruktoseintoleranz)	223100/ 229600	<i>LCT/ALDOB</i>	603202/612724	x		