genetikum

Genliste Carrier Screening

AAAS	ARSB	CLDN19	**DMD**	FANCB	GP1BA	INSR	MKS1	OSTM1	PTH1R	SLC22A5	TNNT1
ABCA12	ARSL	CLN3	DMP1	**FANCC**	GP9	INVS	**MLC1**	**OTC**	PTPRC	SLC25A13	**TNXB**
ABCA3	**ARX**	CLN5	DNAH5	FANCG	GRHPR	IQCB1	MLYCD	P3H1	PTS	SLC25A15	TPO
ABCA4	**ASL**	CLN6	DNAI1	FBLN5	**GRIP1**	ITGA6	MMAA	**PAH**	PUS1	SLC25A20	TPP1
ABCB11	ASNS	CLN8	DNAI2	FBP1	GSS	ITGB4	MMAB	PAX6	PYGL	SLC25A22	TREX1
ABCC8	**ASPA**	**CLRN1**	DNAJC19	FGA	GTF2H5	IVD	**MMACHC**	PAX8	PYGM	**SLC26A2**	TRHR
ABCD1	ASS1	**CNGB3**	DNMT3B	FGB	GUCY2D	IYD	MMADHC	PC	QDPR	SLC26A3	TRIM32
ACAD9	ATIC	COL11A2	DOK7	FGD4	GUSB	JAK3	**MMUT**	PCBD1	RAB23	**SLC26A4**	TRIM37
ACADL	ATM	COL17A1	DOLK	FGFR2	GYS2	KCNJ1	MOCS1	PCCA	RAB27A	SLC2A1	TRMU
ACADM	ATP6V0A2	COL1A2	DPAGT1	FGG	HADH	KCNJ11	MOCS2	PCCB	RAB3GAP1	SLC34A2	TSEN54
ACADS	ATP6V1B1	COL27A1	DPM1	FH	HADHA	KCNQ1	MOGS	**PCDH15**	RAB3GAP2	SLC35A1	TSFM
ACADSB	ATP7A	COL2A1	DPYD	**FKRP**	HADHB	KCNQ2	MPI	PDHB	RAG1	SLC35A3	TSHB
ACADVL	**ATP7B**	COL4A3	DSP	**FKTN**	HAMP	KCTD7	MPL	PDHX	RAG2	SLC35C1	TSHR
ACAT1	ATP8B1	COL4A4	DUOX2	FLNA	HAX1	KRT18	MPV17	PDP1	RAPSN	SLC35D1	TSPYL1
ACE	ATR	COL4A5	DUOXA2	**FMO3**	**HBA1**	KRT8	MPZ	PDSS1	**RARS2**	**SLC37A4**	TTPA
ACOX1	ATRX	**COL7A1**	**DYNC2H1**	**FMR1**	**HBA2**	**L1CAM**	MRPS16	PDSS2	RDH12	SLC39A4	TWNK
ACSF3	AUH	COQ2	DYSF	FOLR1	**HBB**	LAMA2	MRPS22	PEX1	RELN	SLC3A1	TYMP
ADA	AVPR2	COQ8A	EDA	FOXN1	HESX1	LAMA3	MTM1	PEX10	REN	SLC45A2	**TYR**
ADAMTS13	B4GALT1	CPS1	EDN3	FOXP3	**HEXA**	LAMB2	MTR	PEX2	RLBP1	SLC4A1	TYRP1
ADAMTS2	**BBS1**	CPT1A	EDNRB	FRAS1	HEXB	LAMB3	MTRR	PEX6	RMRP	SLC4A11	UBA1
ADAMTSL2	BBS10	**CPT2**	EFEMP2	FREM2	HFE	LAMC2	MTTP	PEX7	**RNASEH2B**	SLC5A5	UBR1
ADGRG1	BBS12	CRB1	EGR2	FUCA1	HGD	LARGE1	MUTYH	PFKM	RPE65	**SLC6A8**	UGT1A1
ADGRV1	**BBS2**	CRLF1	EIF2AK3	**FXN**	HGSNAT	LBR	**MVK**	PHGDH	**RPGR**	SLC7A7	UQCRB
AFF2	BCHE	CRTAP	EIF2B5	**G6PC1**	HIBCH	LCA5	MYO15A	PHKB	RPGRIP1L	SLC7A9	UQCRQ
AGA	BCKDHA	CSTB	**ELP1**	G6PC3	HJV	LDLR	MYO5A	**PKHD1**	**RS1**	SLC9A6	UROS
AGL	**BCKDHB**	CTNS	EMD	**GAA**	HLCS	LDLRAP1	MYO7A	PKLR	RTEL1	SMARCAL1	USH1C
AGPS	BCS1L	CTSC	ENPP1	GALC	HMGCL	LHCGR	**NAGA**	PLA2G6	SACS	**SMN1**	USH1G
AGT	**BLM**	CTSD	EPB42	GALE	HMGCS2	LHX3	NAGLU	PLCE1	SAMHD1	SMN2	**USH2A**
AGTR1	BRIP1	CTSK	EPM2A	GALK1	HOGA1	LIFR	NAGS	PLEC	SBDS	**SMPD1**	VDR
AGXT	BSND	CYBA	ERBB3	GALNS	HPD	LIPA	NBN	PLEKHG5	SC5D	SNAP29	VLDLR
AHI1	**BTD**	CYBB	**ERCC2**	**GALT**	HPRT1	LMBRD1	NDP	PLG	SCNN1A	SP110	VPS13A
AIRE	BTK	**CYP11A1**	ERCC3	GAMT	**HPS1**	LMNA	NDRG1	PLOD1	SCNN1B	SPR	VPS13B
AKR1D1	CA2	CYP11B1	ERCC4	GATA1	**HPS3**	LOXHD1	NDUFAF5	**PLP1**	SCNN1G	SRD5A2	VPS33B
ALDH3A2	CAPN3	CYP11B2	ERCC5	GATM	HPS4	LPL	NDUFS6	**PMM2**	**SCO2**	ST3GAL5	VPS45
ALDH5A1	CASR	CYP17A1	ERCC6	**GBA1**	HSD17B10	**LRP2**	**NEB**	PMP22	SELENON	STAR	VPS53
ALDH7A1	**CBS**	CYP19A1	ERCC8	GBAP	HSD17B3	LRP5	NEU1	PNPO	SEPSECS	STRA6	VRK1
ALDOB	**CC2D2A**	CYP1B1	ESCO2	**GBE1**	HSD17B4	LRPPRC	NEUROG3	**POLG**	SERPINA1	SUCLG1	VSX2
ALG1	**CCDC88C**	**CYP21A2**	ETFA	GCDH	HSD3B2	LYST	NHLRC1	POMGNT1	SFTPA1	SUMF1	WAS
ALG6	CD3D	CYP21A1P	ETFB	GCH1	HSD3B7	MAN2B1	NPC1	POMT1	SFTPB	SUOX	WNT10A
ALMS1	CD3E	**CYP27A1**	ETFDH	GCK	HSPG2	MAPK10	NPC2	POMT2	SGCA	TAFAZZIN	WNT3
ALPL	CD40LG	**CYP27B1**	ETHE1	GCSH	HYAL1	MAT1A	NPHP1	POR	SGCB	TAT	WNT7A
ALS2	CD96	D2HGDH	EVC	GDF5	HYCC1	MCCC1	NPHP3	POU1F1	SGCD	TBCE	WRN
AMACR	CDH23	DBT	**EVC2**	GFM1	HYLS1	**MCCC2**	NPHP4	PPT1	SGCG	TCIRG1	WT1
AMH	**CEP290**	DCLRE1C	EXOSC3	GJA1	IDS	MCEE	**NPHS1**	PQBP1	SGSH	TECPR2	XPA
AMHR2	CERKL	DCX	EYS	GJB1	**IDUA**	**MCOLN1**	NPHS2	PREPL	SH2D1A	**TF**	**XPC**
AMPD1	CFP	DDB2	F11	**GJB2**	IGBP1	**MCPH1**	**NR0B1**	**PRF1**	SIL1	TFR2	ZIC3
AMT	**CFTR**	DDC	F2	GJC2	IGF1	MECP2	NR2E3	PRKAG2	SKIC3	TG	ZMPSTE24
ANO10	CHRNA1	DGUOK	F5	**GLA**	IGHMBP2	MED12	NTRK1	PRKAR1A	SLC12A1	TGM1	ZNF469
ANTXR2	CHRND	DHCR24	**F8**	GLB1	IGSF1	MED17	NUP62	PROC	SLC12A3	TH	
APTX	**CHRNE**	**DHCR7**	**F9**	GLDC	IKBKG	MEFV	OAT	PROP1	SLC12A6	TIMM8A	
AQP2	CHRNG	**DHDDS**	**FAH**	GLE1	IL2RG	MESP2	**OCA2**	PRPS1	SLC16A1	TK2	
AR	CIITA	DKC1	FAM161A	GNE	IL7R	MFSD8	OCRL	PRX	SLC16A2	**TMEM216**	
ARG1	**CLCN1**	**DLD**	FAM20C	**GNPTAB**	INPP5E	MGAT2	OFD1	PSAP	SLC17A5	TMEM67	
ARSA	CLDN1	DLL3	FANCA	GNS	INS	**MID1**	OPA3	PSAT1	**SLC19A3**	TNFRSF11B	

Von ACMG empfohlen:

= Tier 3

= Tier 2

= Tier 1

Tier 4* < 1/200 Trägerfrequenz (inkl. Tier 3)
Gene/Erkrankungen können je nach Labor variieren

Tier 3* ≥ 1/200 Trägerfrequenz (inkl. Tier 2)
inkl. definierte X-chromosomale Erkrankungen

Tier 2* ≥ 1/100 Trägerfrequenz (inkl. Tier 1)

Tier 1* Mukoviszidose + spinale Muskelatrophie (SMA) +
risikobasiertes Screening