Bundesministerium
Arbeit, Soziales, Gesundheit,
Pflege und Konsumentenschutz

ZENTRALE LISTE RINGVERSUCHE
(Ringversuchsregister gem. § 79 Abs. 1 Zi 3 GTG)
Ringversuche zur externen Qualitätssicherung in der molekularen Gendiagnostik

Legende Methoden:

IHC	Immunohistochemistry
FISH	Fluorescence in situ hybridization
NGS	Next generation sequencing
S	Sequencing
RS	RNAseq
RT	RT-PCR
CNV	Copy number variation
MET	Methylation analysis
SB	Southern blot
X	Sonstige, unbestimmt, gemischt

Anbieter:

BVDH	Berufsverband Deutscher Humangenetiker; Deutschland https://www.bvdh-ringversuche.de/
CAP	College of American Pathologists; USA https://www.cap.org/
CF Network	Cystic Fibrosis European Network; Belgien http://cf.eqascheme.org/
EMQN	European Molecular Genetics Quality Network; UK https://www.emqn.org/
EQUALIS	External quality assurance in laboratory medicine in Sweden; Schweden https://www.equalis.se/en/
ESP-EQA	European Society of Pathology - European Quality Assurance Program; Belgien https://www.esp-pathology.org/esp-foundation/eqa-schemes.html
ETRL	Eurotransplant Reference Laboratory; Niederlande http://etrl.eurotransplant.org/cms/index.php
GenQA	Genomics quality assessment; UK https://www.genqa.org/
INSTAND	Institut für Standardisierung und Dokumentation im medizinischen Laboratorium; Deutschland https://www.instand-ev.de/
ÖQUASTA	Österreichische Gesellschaft für Qualitätssicherung und Standardisierung medizinisch-diagnostischer Untersuchungen; Österreich http://www.oequasta.at/de/
QuIP	Qualitätssicherungs-Initiative in der Pathologie; Deutschland https://quip.eu/de_DE/
RCPA QAP	Royal College of Pathologists of Australasia Quality Assurance Programs; Australien https://rcpaqap.com.au/
RfB (DGKL)	Referenzinstitut für Bioanalytik; Deutschland https://www.rfb.bio/
UHKT	Institute of Hematology and Blood Transfusion; Tschechien https://www.uhkt.cz/laboratories/external-proficiency-testing

Stand: 09.04.2025

Gen, Locus		Anbieter & Methoden													
Bezeichnung	OMIM	BVDH	CAP	CF Network	EMQN	EQUALIS	ESP-EQA	ETRL	GenQA	INSTAND	ÖQUASTA	QuIP	RCPA QAP	RfB (DGKL)	UHKT
15q11-q13 Chromosome Region			S, MET		MET, SB					S					
1p/19q			FISH						X						
ABCB1	*171050													S	
ABCD1	*300371				NGS, CNV										
ABL1	*189980		NGS												
ACAD9	* 611103				NGS, CNV										
ACADM (MCAD)	*607008		NGS, S						X, S						
ACADVL	*609575				NGS, CNV										
ACE (DCP1)	*106180									S				S	
ACTA1	* 102610				NGS, CNV										
ACTC1	*102540		S		S, NGS, CNV										
ACTN2	*102573				NGS, S										
ACVR2A	*102581				S										
ADA	*608958				NGS, S										
ADA2	*607575				S										
ADGRG2	*300572														
ADSSL1	*612498				NGS, CNV										
AFG3L2	*604581				NGS, CNV										
AGL	*610860				NGS, CNV										
AGRN	* 103320				NGS, S										
AIP	*605555								X						
AKT1	*164730				S										
ALDOA	*103850				NGS, S										
ALDOB	*612724									S				S	
ALG10B	*603313														
ALG14	*612866				NGS, S										
ALG2	*607905				NGS, S										
ALK	*105590		NGS, IHC				FISH, IHC, S		X, FISH, IHC			S, IHC, X	S, FISH, IHC		
ALPK3	*617608				S, NGS										
ALS2	*606352				NGS, CNV										
AMACR	*604489											IHC, X			
ANCR (Chr. 15q11-q13)	*601623								X	S					
ANG	*105850				NGS, CNV										
ANK2	*106410														
ANO5	*608662				NGS, S										
ANXA11	*602572				NGS, CNV										
APC	*611731		NGS		S, NGS, CNV				X						
APOB	*107730				S, NGS, CNV				X	S				S	
APOE (Apolipoprotein-E)	*107741		S			S				S				S	
APP	*104760				NGS, CNV										
AR (Androgen receptor)	*313700				NGS, CNV				X				S		
ARID1A	*603024												S		
ARSA	*607574				NGS, CNV										
ASAH1	*613468				NGS, S										
ASCC3	*614217				NGS, S										
ASPA	*608034		S												
ASXL1	*612990									NGS			S		
ATM	*607585		NGS		S, NGS, CNV				X				S		
ATN1	*607462								X						
ATP12A2	*610513				NGS, CNV										
ATP1A3	*182350				NGS, CNV										
ATP7B	*606882				S, NGS, CNV					S				S	
ATXN1	*601556		S		S										
ATXN2	*601517		S		S										
ATXN3	*607047		S		S										
ATXN7	*607640		S		S										
AUH	*600529				NGS, CNV										
AZF	*415000									S					
B2M	* 109700												S		
B3GALNT2	* 610194				NGS, S										
B4GAT1	*605517				NGS, S										
BAG3	*603883				NGS, S										
BCHE	*177400													S	
BCL2	*151430		FISH, IHC									IHC	S		
BCL6	*109565											IHC	S		
BCOR	*300485														
BCR-ABL1 p190 fusion			S, RS, FISH										RS		S
BCR-ABL1 p210 fusion			S, RS, FISH										RS		S
BCR-ABL1 e1a2 fusion													RS		
BCR-ABL1 e1a3 fusion													RS		
BCR-ABL1 e13a2 fusion													RS		
BCR-ABL1 e13a3 fusion													RS		
BCR-ABL1 e14a2 fusion													RS		
BCR-ABL1 e14a3 fusion													RS		
BICD2	*609797				NGS, S										
BIN1	*601248				NGS, S										
BIRC3	*601721												S		
BLM (RECQL3)	*604610		S						X						
BMPR2	*600799														
BRAF	*164757		S, NGS, IHC		S, NGS		IHC, S		X	S		S	S, IHC	S	
BRCA1	*113705		S		S, NGS, CNV				X	S		S			
BRCA2	*600185		S		S, NGS, CNV				X	S		S			
BRIP1	*605882														
BTBD7 (FUP1)	*610386				S										
BTK	*300300								X	S			S		
C19ORF12	*614297				NGS, CNV										
C9orf72	*614260				NGS, CNV				X						
CACNA1A	*601011		S		S										
CACNA1C	*114205				NGS, S										
CACNA1G	* 604065				NGS, CNV										
CACNA1S	*114208				NGS, S										
CACNB2	*600003														
CALM1	* 114180				NGS, S										
CALM2	*114182				NGS, S										
CALM3	*114183				NGS, S										
CALR	*109091		S, RS, NGS							S			S		
CAPN3	*114240				NGS, S										

CARD11	***607210**											S			
CASQ2	***114251**				NGS, S										
CASP10	***601762**														
CAV3	***601253**				NGS, S										
CAVIN1	***603198**				NGS, S										
CBFB-MYH11 fusion			S, RS												
CBL	***165360**											S			
CCDC78	***614666**				NGS, S										
CCND1 (Cyclin D1)	*** 168461**										IHC				
CCNF	***600227**				NGS, CNV										
CCR5	***601373**									S		IHC	S		
CD117			IHC								IHC	IHC			
CD20			IHC								IHC				
CD3											IHC				
CD30			IHC								IHC				
CD4	***186940**											IHC			
CD5											IHC				
CD74-ROS1 fusion			RS												
CDC73	***607393**								X						
CD79B	***147245**											S			
CDH1	***192090**											IHC			
CDK12	***615514**														
CDKN1B (KIP1)	*** 600778**								X						
CDKN1C (KIP2)	***600856**				CNV, MET										
CDKN2A	***600160**		S, NGS								S	S			
CDKN2B	***600431**		NGS												
CEBPA	***116897**									NGS		S			
CETP	***118470**												S		
CFL2	***601443**				NGS, S										
CFTR	***602421**		S	S	S				X	S, NGS					
CHAT	***118490**				NGS, S										
CHCHD10	***615903**				NGS, CNV										
CHCHD2	***616244**														
CHEK2	***604373**		S		S, NGS, CNV										
CHGA	*** 118910**										IHC				
CHKB	***612395**				NGS, S										
CHMP2B	***609512**				NGS, CNV										
CHRNA1	***100690**				NGS, S										
CHRNB	***100710**				NGS, S										
CHRND	***100720**				NGS, S										
CHRNE	***100725**				NGS, S										
CHRNG	***100730**				NGS, S										
CLCN2	***600570**				NGS, CNV										
CLN6	***606725**				NGS, CNV										
CMET	***164860**						IHC, FISH								
CNBP	***116955**				S, SB										
COASY	***609855**				NGS, CNV										
COL1A1	***120150**				S, NGS, CNV								S		
COL1A1-PDFGB fusion			RT												
COL1A2	***120160**				S, NGS, CNV										
COL12A1	***120320**				NGS, S										
COL13A1	***120350**				NGS, S										
COL2A1	***120140**				S, NGS, CNV				X						
COL4A3	***120070**														
COL4A4	***120131**														
COL4A5	***303630**								X						
COL6A1	***120220**				NGS, S										
COL6A2	***120240**				NGS, S										
COL6A3	***120250**				NGS, S										
COL9A1	***120210**				S, NGS, CNV										
COL9A2	***120260**				S, NGS, CNV										
COL11A1	***120280**				S, NGS, CNV										
COL11A2	***120290**				S, NGS, CNV										
COLQ	***603033**				NGS, S										
COMT	***116790**		S												
COX6A2	***602009**				NGS, S										
CP	***117700**				NGS, CNV										
CPT2	***600650**				NGS, S										
CREBBP	***600140**											S			
CRPPA (ISPD)	***614631**				NGS, S										
CRYAB	***123590**				NGS, S										
CSF1R	***164770**				NGS										
CSF3R	***138971**											S			
CSRP3	***600824**				S, NGS										
CTNNB1 (Beta-catenin)	***116806**														
CTSF	***603539**				NGS, CNV										
CXCR4	*** 162643**											S			
CYP21A2	***613815**				S, NGS, SB, CNV					S					
CYP27A1	***606530**				NGS, CNV										
CYP2B6 (Cytochrome P450)	***123930**		S		S, NGS				X				S		
CYP2C19 (Cytochrome P450)	***124020**		S		S, NGS, CNV				X	S			S		
CYP2C8	***601129**												S		
CYP2C9	***601130**		S		S, NGS, CNV				X	S			S		
CYP2D6 (Cytochrome P450)	***124030**		S		S, NGS, CNV				X	S			S		
CYP3A4 (Cytochrome P450)	***124010**		S		S, NGS, CNV								S		
CYP3A5 (Cytochrome P450)	***605325**		S		S, NGS, CNV				X				S		
CYP4F2	***604426**		S												
CYP7B1	*** 603711**				NGS, CNV										
D4Z4 microsatellite repeat (4q35)					S										
DAG1	***128239**				NGS, S										
DARS2	***610956**				NGS, CNV										
DCLRE1C	***605988**				NGS, S										
DCTN1	***601143**				NGS, CNV										
DDIT3	***126337**								X						
DDR2	***191311**						S								
DES	***125660**				NGS, S										
DIDO1	***604140**				S										
DMD (Dystrophin)	***300377**		S		S, CNV				X	S					
DMPK	***605377**		S		S, SB				X						

DNAJB6	***611332**				NGS, S										
DNAJC5	***611203**				NGS, CNV										
DNAJC6	***608375**				NGS, CNV										
DNM2	***602378**				NGS, S										
DNMT1	***126375**				NGS, CNV										
DNMT3A	***602769**												S		
DOK7	***610285**				NGS, S										
DOLK	***610746**				NGS, S										
DPAGT1	***191350**				NGS, S										
DPM2	***603564**				NGS, S										
DPYD (DPD)	***612779**		S		S, NGS, CNV				X	S				S	
DYNC1H1	***600112**				NGS, S										
DYSF	***603009**				NGS, S										
ECEL1	***605896**				NGS, S										
EGFR	***131550**		NGS		S, NGS		IHC, FISH		X			S	S	S	
EIF2B1	***606686**				NGS, CNV										
EIF2B2	***606454**				NGS, CNV										
EIF2B3	***606273**				NGS, CNV										
EIF2B4	***606687**				NGS, CNV										
EIF2B5	***603945**				NGS, CNV										
ELOVL4	***605512**				NGS, CNV										
ELP1	***603722**		S												
EMD	***300384**				NGS, S										
EML4-ALK fusion			RS												
ENO3	***131370**				NGS, S										
EP300 (p300)	***602700**												S		
EPCAM	***185535**				S, NGS										
EPG5	***615068**				S, NGS										
EPM2A	***607566**				NGS, CNV										
ERBB2 (HER2)	***164870**		NGS, FISH		S, NGS				S			IHC, X	IHC, X		
ERBB4	***600543**														
ESR1 (ER)	***133430**		IHC		S, CNV										
ETFA	***608053**				S, CNV										
ETFB	***130410**				S, CNV										
ETFDH	*** 231675**				S, CNV										
ETV6	***600618**		NGS										S, IHC, FISH		
ETV6-NTRK3 fusion			RT, RS												
EWSR1	***133450**		FISH						X						
EWSR1-ATF1 fusion			RT												
EWSR1-ERG fusion			RT												
EWSR1-FLI1 fusion			RT												
EWSR1-WT1 fusion			RT												
EXOSC3	*** 606489**				NGS, S										
EZH2	***601573**												S		
F12 (Faktor 12)	***610619**													S	
F13A1 (Faktor 13, A1 subunit)	***134570**													S	
F2 (Faktor II, Prothrombin)	***176930**		S			S				S			S	S	
F5 (Faktor V, Leiden, fVL)	***612309**		S		S, NGS	S				S			S	S	
F7 (Faktor 7)	***613878**													S	
FANCC	***613899**		S												
FBN1	***134797**														
FBXO7	***605648**				NGS, CNV										
FBXW7	***606278**														
FDX2	***614585**				NGS, S										
FECH	***612386**				S										
FGB (Fibrinogen beta)	***134830**													S	
FGFR1	***136350**						FISH		X						
FGFR1-FGFR2 fusion					S										
FGFR2	***176943**				S				X						
FGFR3	***134934**		NGS		S				X			S			
FGFR3-TACC3 fusion			RS												
FGFR4	*** 134935**				S										
FH	***136850**								X						
FHL1	***300163**				S, NGS										
FHOD3	***609691**				S, NGS										
FIG4	***609390**				NGS, CNV										
FKBP14	***614505**				NGS, S										
FKRP	***606596**				NGS, S										
FKTN	***607440**				NGS, S										
FLCN (Folliculin)	***607273**														
FLNC	***102565**				S, NGS										
FLT3	***136351**		NGS							NGS			S		
FLT3-ITD fusion			S, RS										S		
FLT3-TKD fusion			S, RS										S		
FMR1 (FRAXA)	***309550**		S		S, MET, SB, X				X	S					
FOXO1	***136533**		FISH						X				S		
FTL	***134790**				NGS, CNV										
FUS	***137070**		FISH		NGS, CNV				X						
FUS-DDIT3 fusion			RT												
FXN	***606829**		S		S, SB										
FXR1	***600819**				NGS, S										
G6PC	***613742**		S												
G6PD	***305900**		S												
GAA	***606800**				NGS, S										
GALT	*** 606999**								X						
GARS1	***600287**				NGS, S										
GATA2	***137295**												S		
GBA	***606463**		S												
GBE1	***607839**				NGS, S										
GCH1	***600225**				NGS, CNV										
GCK (MODY2)	***138079**				S, NGS, CNV										
GFAP	***137780**				NGS, CNV										
GFPT1	***138292**				NGS, S										
GGPS1	***606982**				NGS, S										
GJB1 (CX32)	***304040**				S, NGS, CNV				X						
GJB2 (CNX26) (Connexin 26)	***121011**		S		S, NGS, CNV					S					
GJB6 (CX30)	***604418**				S, NGS, CNV										
GLA (Galactosidase)	***300644**				S, NGS, CNV				X						
GMPPB	***615320**				NGS, S										

GNA11	*139313														
GNAQ	*600998														
GNAS	*139320														
GNB5	*604447				NGS, S										
GNE	*603824				NGS, S										
GPD1L	*611778														
GRN	*138945				NGS, CNV										
GYG1	* 603942				NGS, S										
GYS1	* 138570				NGS, S										
H19/IGF2 imprinting control region	*616186														
H3F3A	*601128														
HABP2 (FSAP)	*603924													S	
HACD1	*610467				NGS, S										
HADHA	*600890				NGS, S										
HADHB	*143450				NGS, S										
HAVCR2	*606652												S		
HBA1	*141800									S			S		
HBA2	*141850									S			S		
HBB (Hämoglobin beta)	*141900									S			S		
HCN4	*605206														
HER2	*164870		FISH, IHC				IHC, FISH		X			IHC, X	IHC, X		
HEXA	*606869		S		NGS, CNV										
HEXB	*606873				NGS, CNV										
HFE	*613609		S		S, NGS	S				S			S	S	
HLA-A	*142800		S					X		S, X					
HLA-B	*142830		S		S, NGS, CNV			X	X	S, X			S	S	S
HLA-C	*142840		S					X		S, X					
HLA-DP alleles			X							S					
HLA-DQ alleles			X							S, X			S		S
HLA-DR alleles			X					X		S, X					
HMBS	*609806				S, NGS, CNV										
HNF1A (MODY3)	*142410				S, NGS, CNV										
HNF1B (MODY5)	*189907				S, NGS, CNV										
HNF4A (MODY1)	*600281				S, NGS, CNV										
HNRNPA1	*164017				NGS, CNV										
HNRNPA2B1	* 600124				S, NGS										
HNRNPDL	*607137				NGS, S										
HPV DNA										S		S	S		
HRAS	*190020								NGS						
HSPB1	*602195				NGS, S										
HSPB8	*608014				NGS, S										
HTRA1	*602194				NGS, CNV										
HTT (Huntingtin)	*613004		S		S				X	S					
HUWE1	*300697														
ICR1	*616186				S, Met, CNV										
ID3	*600277												S		
IDH1	*147700		NGS, X						X	NGS		S	S, IHC		
IDH2	*147650		NGS, X		S, NGS				X	NGS		S	S		
IFNL3 (IL28B)	*607402		S							S				S	
IFNL4	*615090													S	
IGH	*147070		S, FISH										S		
IGH-BCL2 major fusion			S												
IGH-BCL2 minor fusion			S												
IGH-CCND1 fusion			S												
IGHMBP2	*600502				NGS, S										
IGHV	*147070		S						X			S			
IGK (Ig Kappa)			S, ISH												
IG light chain												S			
IKBKAP (ELP1)	*603722		S												
IKZF2	*606234												S		
IL6	*147620													S	
INPP5K	*607875				NGS, S										
INSM1	*600010												IHC		
IRF4 (MUM1)	*601900												IHC		
ISCU	*611911				NGS, S										
ITGA2 (GPIa/IIa)	*192974									S				S	
ITGA7	*600536				NGS, S										
ITGB3 (GP3A, HP 1a/1b)	*173470									S				S	
ITM2B	*603904				NGS, CNV										
JAG2	*602570				S, NGS										
JAK1													S		
JAK2	*147796		S, RS, NGS		S, NGS	S				S			S		
JAK3	*600173												S		
KBTBD13	*613727				NGS, S										
KCNC3	*176264				NGS, CNV										
KCND3	*605411				NGS, CNV										
KCNE1	*176261				S, NGS, CNV										
KCNE2	*603796														
KCNE3	*604433														
KCNH2	*152427				S, NGS, CNV										
KCNJ2	*600681				NGS, S										
KCNQ1	*607542				S, NGS, CNV										
KCNQ1OT1 (LIT1)	*604115				CNV, MET										
KDR	*191306														
KEAP1	* 606016								X						
KI-67	*176741		IHC									IHC, X			
KIF5A	*602821				NGS, CNV										
KIT	*164920		NGS		S, NGS				X	S		S	S	S	
KLF2	*602016												S		
KLHL40	*615340				NGS, S										
KLHL41	*607701				NGS, S										
KLMT2A	*159555												S		
KMT2D	*602113														
KRAS	*190070		NGS		S, NGS		S		X	S		S	S	S	
KRT7	*148059											IHC, X			
KRT20	*608218											IHC, X			
LAMA2	*156225				NGS, S										
LAMP2	*309060				NGS, S										
LARGE1	*603590				NGS, S										

LCT (Lactase)	*603202					S				S				S	
LDB3	*605906				NGS, S										
LDHA	*150000				NGS, S										
LDLR	*606945				S, NGS, CNV				X						
LMNA	*150330				NGS, S										
LMOD3	*616112				NGS, S										
LPIN1	*605518				NGS, S										
LRP4	*604270				NGS, S										
LRRK2	*609007				NGS, CNV										
LYST	*606897				NGS, CNV										
MALT1	*604860		FISH												
MAP3K20	*609479				NGS, S										
MAPT	*157140				NGS, CNV										
MATR3	*164015				NGS, S										
MAX	*154950								X						
MCOLN1	*605248		S												
MDM2	*164785								X						
MECP2	*300005		S						X						
MEF2B	*600661												S		
MEFV	*608107				S, NGS, CNV										
MEGF10	*612453				NGS, S										
MEN1	*613733								X						
MET	*164860		NGS		S, NGS				X, FISH, IHC			S, X			
MFN2	*608507								S						
MGMT	*156569		X						MET			S	S, MET		
MICU1					NGS, S										
MLH1	*120436		S, NGS, IHC		S, NGS, CNV				MET, X	S		IHC, MET	IHC		
MLL-PTD (KMT2A-PTD) fusion			S, RS												
MPL	*159530		S, RS, NGS							S			S		
MPZ (MPP, P0)	*159440				S, NGS, CNV				X						
MRE11	*600814				S										
MSH1															
MSH2	*609309		S, NGS, IHC		S, NGS, CNV				X	S		IHC			
MSH6	*600678		S, NGS, IHC		S, NGS, CNV				X	S		IHC			
MSTO1	* 617619				NGS, S										
MTHFR	*607093		S			S				S				S	
MTM1	*300415				NGS, S										
MTRNR1 (MT-RNR1)	* 561000								X						
MUSK	*601296				NGS, S										
MUTYH	*604933				S, NGS, CNV				X						
MVK	*251170				S, NGS, CNV										
MYBPC1	*160794				NGS, S										
MYBPC3	*600958		S		S, NGS, CNV										
MYC	*190080		NGS, IHC										S		
MYCN / NMYC	*164840		NGS												
MYD88	*612260									S			S		
MYH2	*160740				NGS, S										
MYH3	*160720				NGS, S										
MYH7	*160760		S		S, NGS, CNV										
MYL1	*160780				NGS, S										
MYL2	*160781		S		S, NGS, CNV										
MYL3	*160790		S		S, NGS, CNV										
MYMK	*615345				NGS, S										
MYO18B	*607295				NGS, S										
MYO9A	*604875				NGS, S										
MYOD1	*159970				NGS, S										
MYOG	*159980				S, NGS, CNV							IHC			
MYORG	*618255				NGS, CNV										
MYOT	*604103				NGS, S										
MYPN	*608517				NGS, S										
NEB	*161650				NGS, S										
NEFH	*162230														
NHLRC1	*608072				NGS, CNV										
NFKBIE	*604548												S		
NKX3-1	*602041											IHC, X			
NLRP3	*606416				S, NGS, CNV										
NOD2	*605956				S, NGS, CNV									S	
NOTCH1	*190198												S		
NOTCH3	*600276				NGS, CNV										
NPC1	*607623				NGS, CNV										
NPC2	*601015				NGS, CNV										
NPM1	*164040		S, RS, NGS							NGS			S		
NR4A3	*600542								X						
NRAS	*164790		NGS		S, NGS		S		NGS			S	S	S	
NTHL1	* 602656								X						
NTRK1	*191315						FISH, S		NGS, FISH				S, IHC, FISH		
NTRK2	*600456								NGS, FISH				S, IHC, FISH		
NTRK3	*191316								NGS, FISH				S, IHC, FISH		
NUDT15	*615792		S		S, NGS, CNV				X						
OPTN	* 602432				NGS, CNV										
OPRM1	*600018		S												
ORAI1	*610277				NGS, S										
p16			IHC												
p53	*602704		IHC						X						
PAH	*612349				S										
PALB2	*610355		S		S, NGS, CNV				X						
PANK2	*606157				NGS, CNV										
PARK7	#606324				NGS, CNV										
PAX3	* 606597												S, IHC, FISH		
PAX3-FOXO1 fusion			RT												
PAX7	*167410				NGS, S										
PAX7-FOXO1 fusion			RT												
PAX8-PPARG fusion			RS												
PAX8	*167415		IHC												
PCSK9	*607786				S, NGS, CNV										
PDCD1 (PD1)	*600244												IHC		
PDCD1LG2	* 605723												S		
PDGFB	* 190040				NGS, CNV										
PDGFRA	*173490		NGS						X			S			

PDGFRB	*173410				NGS, CNV										
PD-L1 (CD274)	*605402		IHC									IHC	IHC , S		
PFKM	*610681				NGS, S										
PFN1	*176610				NGS, CNV										
PGAM2	*612931				NGS, S										
PHOX2B	*603851				S, NGS										
PIK3CA	*171834		NGS		S, NGS, CNV		S		X			S			
PIM1	*164960												S		
PINK1	*608309				NGS, CNV										
PKD1 (Polycystin 1)	*601313				S, NGS, CNV				X						
PKD2 (Polycystin 2)	*173910				S, NGS, CNV				X						
PLA2G6	* 603604				NGS, CNV										
PLN	*172405				S, NGS										
PML-RARA fusion			S, RS										RS		
PMP22 (GAS3)	*601097				S, NGS, CNV				X						
PMS2	*600259		S, NGS, IHC		S, NGS, CNV				X	S		IHC	IHC		
POLD1	* 174761								X						
POLE (DNA Polymerase Epsilon)	* 174762								S						
POLG	*174763								X						
POLG2	*604983				NGS, S										
POMGNT1	*606822				NGS, S										
POMGNT2	*614828				NGS, S										
POMK	* 615247				NGS, S										
POMT1	* 607423				NGS, S										
POMT2	* 607439				NGS, S										
PPOX	*600923				S, NGS, CNV										
PRF1	*170280														
PRKAG2	*602743				S, NGS										
PRKN	*602544				NGS, CNV										
PRNP (PRP, PRIP)	*176640				NGS, CNV										
PROC (PC, Protein C)	*612283									S					
PROS1 (Protein S)	*176880						IHC, FISH			S					
PRPH	*170710														
PSAP	*176801											IHC, X			
PSEN1	*104311				NGS, CNV										
PSEN2	*600759				NGS, CNV										
PSTPIP1	*606347				S										
PTEN	*601728		NGS		S, NGS, CNV		IHC		X				S		
PTPN11	*176876								X						
PTPRD	*601598												S		
PYGM	*608455				NGS, S										
PYROXD1	*617220				NGS, S										
Quasimonomorphic mononucleotide repeats					S										
RAD51C	* 602774				S, NGS, CNV				X						
RAD51D	* 602954				S, NGS, CNV				X						
RAD54B	*604289														
RAD54L	*603615														
RAG1	*179615				NGS, S										
RAG2	*179616				NGS, S										
RAPSN	*601592				NGS, S										
RB1	*614041				S, NGS, CNV										
RBCK1	* 610924				NGS, S										
RET	*164761		S, NGS		S, NGS, CNV				X, FISH, IHC	S		S			
RHD (Rhesus blood group, D antigen)			S, X												
RHOA	*165390												S		
RNF216	*609948				NGS, CNV										
ROS1	*165020						FISH, IHC		X, FISH, IHC			S, IHC, X	S, IHC, FISH		
RPE65	*180069				S, NGS										
RPGR	*312610				S, NGS										
RUNX1	* 151385									NGS			S		
RUNX1-RUNX1T1 fusion			S, RS							NGS					
RXYLT1 (TMEM5)	*605862				NGS, S										
RYR1	*180901				S, NGS, CNV										
RYR2	*180902				NGS, S										
RYR3	*180903				S										
SCN1A	*182389								X						
SCN1B	*600235														
SCN3B	*608214														
SCN4A	*603967				NGS, S										
SCN5A	*600163				S, NGS, CNV										
SDC1 (CD138)	*186355												IHC		
SDHA	*600857								X						
SDHAF2	*613019								X						
SDHB	*185470								X						
SDHC	*602413								X						
SDHD	*602690								X						
SEC31A	*610257				S										
SELENON	*606210				NGS, S										
SERPINA1 (AAT; PI)	*107400		S							S				S	
SERPINC1 (Antithrombin, AT3)	*107300									S				S	
SERPINE1 (PAI1)	*173360		S							S				S	
SETBP1	*611060												S		
SETX	*608465				NGS, CNV										
SF3B1	*605590									S			S		
SGCA	*600119														
SGCB	* 600900				NGS, S										
SGCD	* 601411				NGS, S										
SGCG	*608896				NGS, S										
SHOX	*312865				S, NGS, CNV										
SIL1	* 608005				NGS, S										
SLC18A3	* 600336				NGS, S										
SLC20A2	*158378				NGS, CNV										
SLC22A5	*603377				NGS, S										
SLC25A1	*190315				NGS, S										
SLC25A4	*103220				NGS, S										
SLC45A3	*605097											IHC, X			
SLC45A3-BRAF fusion			RS												
SLC52A2	*607882				NGS, S										
SLC52A3	*613350				NGS, S										

SLC5A7	***608761**				NGS, S										
SLCO1B1	***604843**		S		S, NGS, CNV				X						
SMAD4	***600993**														
SMARCB1	***601607**														
SMCHD1	***614982**				S, MET, CNV										
SMN1	***600354**		S		S				S	S					
SMN2	***601627**		S						S	S					
SMO	***601500**														
SMPD1	***607608**		S												
SNCA	***163890**				NGS, CNV										
SNRPN	***182279**								X	S					
SOCS1	***603597**												S		
SOD1	***147450**				S, NGS, CNV										
SOX10	*** 602229**												IHC		
SOX11	***600898**												IHC		
SOX9	***608160**								X						
SPEG	***615950**				NGS, S										
SPRED1	*** 609291**								X						
SPAST	*** 604277**				NGS, CNV										
SPG11	*** 610844**				NGS, CNV										
SPTBN2	***604985**														
SQSTM1	***601530**				NGS, CNV										
SRC	***190090**														
SRSF2	***600813**												S		
SRY	*** 480000**								X						
SS18	***600192**								X						
SS18-SSX1 fusion			RT									IHC			
SS18-SSX2 fusion			RT									IHC			
STAC3	*** 615521**				NGS, S										
STAG2	***300826**												S		
STAT3	***102582**									S			S		
STAT5B	*** 604260**												S		
STAT6	***601512**											IHC	S		
STIM1	***605921**				NGS, S										
STK11	***602216**								X						
SUCLA2	***603921**				NGS, S										
SULF2	***610013**				S										
SYNE1	***608441**				NGS, S										
SYNE2	***608442**				NGS, S										
SYNJ1	*** 604297**				NGS, CNV										
SYP	*** 313475**											IHC			
SYT2	*** 600104**				NGS, S										
TANGO2	***616830**				NGS, S										
TARDBP	*** 605078**				NGS, CNV										
TBK1	*** 604834**				NGS, CNV										
TBX1	***602054**														
TCAP	***604488**				NGS, S										
TCF3	***147141**												S		
TECRL	*** 617242**				NGS, S										
TERT	***187270**								X						
TET2	***612839**												S		
TFE3	*** 314310**								X						
TIA1	***603518**				NGS, S										
TK2	***188250**				NGS, S										
TMEM127	***613403**								X						
TMEM240	***616101**				NGS, CNV										
TNFA	***191160**													S	
TNFAIP3	***191163**				S								S		
TNFRSF1A	***191190**				S, NGS, CNV										
TNNC1 (Troponin C, TNC)	***191040**				NGS, S										
TNNC2	***191039**				NGS, S										
TNNI2	***191043**				NGS, S										
TNNI3	***191044**		S		S, NGS, CNV										
TNNT1	***191041**				NGS, S										
TNNT2	***191045**		S		S, NGS, CNV										
TNNT3	***600692**				NGS, S										
TNPO3	***610032**				NGS, S										
TOR1AIP1	***614512**				NGS, S										
TP53	***191170**		NGS		S, NGS, CNV				X	S, NGS			S, FISH		
TPM1	***191010**		S		S, NGS, CNV										
TPM2	***190990**				NGS, S										
TPM3	***191030**				NGS, S										
TPMT	***187680**		S		S, NGS, CNV				X	S				S	
TRAPPC11	***614138**				NGS, S										
TRB (TC receptor beta)			S									S	S		
TRDN	***603283**				NGS, S										
TREM2	***605086**				NGS, CNV										
TRG (TC receptor gamma)			S									S	S		
TRIM32 / HT2A / BBS11	***602290**				NGS, S										
TRIM63 / RNF28 / SMRZ / MURF1	***606131**				NGS, S										
TRIP4	*** 604501**				NGS, S										
TSC1	***605284**								X						
TSC2	***191092**								X						
TTC19	***613814**				NGS, CNV										
TTN	***188840**				NGS, S										
TTR (TBPA, PALB)	***176300**				NGS, S										
TYMP	***131222**				NGS, S										
TYROBP	*** 604142**				NGS, CNV										
TRPV4	*** 605427**				NGS, S										
TSFM	*** 604723**				NGS, S										
U2AF1	***191317**												S		
UBA1	***314370**				NGS, S										
UBQLN2	***300264**				NGS, CNV										
UGT1A1	***191740**		S		S, NGS, CNV				X	S				S	
UPD11 (Uniparental Disomy 11)					S, CNV				X						
UPD14 (Uniparental Disomy 14)									X						
UPD15 (Uniparental Disomy 15)									X						
UPD7 (Uniparental Disomy 7)					S, CNV				X						
UROD	***613521**				S, NGS										

UROS	***606938**				S, NGS										
USP6	***604334**								X						
VAMP1	***185880**				NGS, S										
VAPB	***605704**				NGS, CNV										
VCP	***601023**				NGS, CNV										
VDR	***601769**													S	
VHL	***608537**				S, NGS, CNV				X						
VKORC1 (Vitamin K-Epoxid Reductase)	***608547**		S		S, NGS, CNV				X	S				S	
VMA21	***300913**				NGS, S										
VPS13A	***605978**				NGS, CNV										
VPS35	***601501**				NGS, CNV										
VRK1	***602168**				NGS, S										
WDR45	*** 300526**				NGS, CNV										
WT1	***607102**		NGS										S		
X-linked genes									X						
XPO1	***602559**												S		
XPR1	***605237**				NGS, CNV										
Y-chromosome microdeletions					X					S					
ZNF346 , JAZ	*** 605308**								X						
ZRSR2	***300028**														

Krankheit, Syndrom, Phänotyp
Bezeichnung
3-methylglutaconic aciduria, type I
Abacavir Hypersensitivität
ACADM deficiency (Acyl-CoA dehydrogenase, medium-chain deficiency; MCAD deficiency)
Achondrogenesis
Achondroplasia
Acne inversa, familial, 3
Acute Lymphoblastic Leukaemia (ALL)
Acute Myeloid Leukemia (AML)
Adenosine deaminase deficiency, partial
Adrenoleukodystrophy (ALD)
Adrenomyeloneuropathy (AMN)
Agammaglobulinemia
Albinism
Alexander disease
Alpha-1-Antitrypsin (AAT) Mangel
Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunit
Alport syndrome
Alternating hemiplegia of childhood 2
Alzheimer disease 2 (AD2; late onset)
Aminoglycoside-induced deafness
Amyotrophic lateral sclerosis (ALS)
Amyotrophic lateral sclerosis 2, juvenile (ALS2)
Andersen syndrome (Andersen cardiodysrhythmic periodic paralysis; long QT syndrome 7)
Androgen Insensitivity syndrome
Aneuploidy / Chromosomal rearrangements
Angelman-Syndrom
Antithrombin III deficiency
Apert syndrome
Aphasia, primary progressive
Arthrogryposis, distal, various types
Arthrogryposis, distal, type 2A (Freeman-Sheldon)
Arthrogryposis, distal, type 2B3 (Sheldon-Hall)
Arthrogryposis, distal, with impaired proprioception and touch
Arthrogryposis multiplex congenita 3, myogenic type
Arthrogryposis multiplex congenita 6
Ataxia telangiecstasia (ATM, Louis-Bar syndrome)
Ataxia-telangiecstasia-like disorder 1 (ATLD1)
Ataxias and Hereditary Spastic Paraplegia (HSP) / Friedreich Ataxia / Spinocerebellar ataxia (SCA)
Autoimmune disease, multisystem, infantile-onset, 1
Autosomal dominant polycystic kidney disease (ADPKD)
Avascular necrosis of femoral head, primary, 2
Bardet-Biedl syndrome 11
Basal ganglia calcification, idiopathic
Becker Muscular Dystrophy (BMD)
Beckwith-Wiedemann Syndrom (BWS)
Bethlem myopathy 1

Bethlem myopathy 2
Bile acid synthesis defect, congenital, 3
Birdshot chorioretinopathy
Birt Hogg Dube Syndrome (BHD)
Blau syndrome
Blasenkarzinom (Bladder cancer)
Bleeding disorder, platelet-type, 16 (BDPLT16)
Bleeding disorder, platelet-type, 24 (BDPLT24)
Bleeding disorder, platelet-type, 9 (BDPLT9)
Bloom Syndrom
Bosma arhinia microphthalmia syndrome (BAMS)
Brachyolmia type 3
Breast/ovarian cancer, familial (*BRCA*) (HBOC)
Bronchiectasis
Brown-Vialetto-Van Laere syndrome
Brugada syndrome
Butyrylcholinesterase deficiency
Canavan disease
CAPOS syndrome
CARASIL syndrome
Carbamazepin Hypersensitivität
Cardiac arrythmias
Cardiomyopathies (sequencing panel)
Cardiomyopathy, hypertrophic
Carey-Fineman-Ziter syndrome
Carnitine deficiency, systemic primary
Carpal Tunnel syndrome CTS1
Cataract, multiple types
Catecholaminergic polymorphic ventricular tachycardia (CPVT)
Cenani-Lenz syndactyly syndrome
Central core disease
Central hypoventilation syndrome,congenital, 1, CCHS1
Centronuclear myopathy
Cerebellar ataxia
Cerebellar ataxia and hypogonadotropic hypogonadism
Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant
Cerebral amyloid angiopathy, PRNP-related
Cerebral arteriopathy, with subcortical infarcts and leukoencephalopathy
Cerebrotendinous xanthomatosis (CTX)
Ceroid lipofuscinosis, neuronal, 11
Ceroid lipofuscinosis, neuronal, 13 (Kufs type)
Ceroid lipofuscinosis, neuronal, 4 (Kufs type)
Ceroid lipofuscinosis, neuronal, 6A
Ceroid lipofuscinosis, neuronal, 6B (Kufs type)
Charcot-Marie-Tooth disease / Hereditary Liability to Pressure Palsies / Hereditary Motor and Sensory Neuropathy
Chediak-Higashi syndrome
Cholangiocarcinoma (bile duct cancer)
Chorea Huntington

Choreoacanthocytosis
Chorionic Villus Sampling (CVS)
Creatine phosphokinase, elevated serum
Chromosome breakage syndromes
Chronic Lymphocytic Leukaemia (CLL), Myelodysplastic syndrome (MDS)
Chronic Myeloid Leukaemia (CML)
Coeliac disease susceptibility (Zöliakie)
Colorectal cancer (CRC)
Combined cellular and humoral immune defects with granulomas
Combined D-2- and L-2-hydroxyglutaric aciduria
Combined deficiency of vitamin K-dependent clotting factors
Combined oxidative phosphorylation deficiency 3
Congenital absence of the vas deferens (bilateral, X-linked)
Congenital adrenal hyperplasia (Adrenogenitales Syndrom)
Congenital afibrinogenemia
Congenital disorder of glycosylation
Congenital dysfibrinogenemia
Congenital hypotonia, epilepsy, developmental delay, digital anomalies
Congenital lactase deficiency
Congenital muscular dystrophies
Congenital myopathy/myopathies
Congenital myopathy 18 due to dihydropyridine receptor defect
Congenital myopathy 3 with rigid spine
Congenital myopathy 6 with ophthalmoplegia
Contractures, pterygia, and spondylocarpostarsal fusion syndrome
Cornelia de Lange syndrome
Cortical dysplasia, complex, with other brain malformations 13
Coumarin/Warfarin Resistenz/Sensitivität
Cowden syndrome
CPT2 II deficiency (various)
Creutzfeldt-Jakob disease
Crouzon Syndrome
Cutaneous mastocytosis
Cystic fibrosis (CF; Mukoviszidose)
Danon disease
Dapsone Hypersensitivität Syndrom (DHS)
Dementia
Dentatorubral-pallidoluysian atrophy (DRPLA)
Dermatofibrosarcoma protuberans
Developmental and epileptic encephalopathy
Diarrhea 5, with tufting enteropathy, congenital (DIAR5)
Di-George syndrome
Digital arthropathy-brachydactyly, familial
Dihydropyrimidin-Dehydrogenase Mangel (5-Fluorouracil Toxizität)
Dravet syndrome
Duchenne/Becker Muscular Dystrophy
Dystonia 12 (DYT12)
Dystonia 27

Dystonia, DOPA-responsive
Dystransthyretinemic hyperthyroxinemia DTTRH
Ehlers Danlos Syndrome
Emery-Dreifuss muscular dystrophies (EDMD)
Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8
Encephalopathy, acute, infection-induced, 4, susceptibility to
Endometrial cancer
Epidermolysis bullosa simplex 5
Epilepsy, progressive myoclonic 2A (Lafora)
Epilepsy, progressive myoclonic 2B (Lafora)
Epilepsy, susceptibility to
Escobar syndrome
Estrogen resistance / insensitivity
Fabry disease
Farber lipogranulomatosis
Factor VII deficiency
Factor XII deficiency
Factor XIIIA deficiency
Faktor V deficiency
Familial adenomatous polyposis (FAP1, APC, FPC)
Familial atrial fibrillation (ATFB9)
Familial endocrine tumour predisposition disorders
Familial hypercholesterolemia 1
Familial hypercholesterolemia 2 (Familial defective apolipoprotein B-100)
Familial hypercholesterolemia 3
Familial hypoparathyroidism
Familial mediterranean fever (=hereditary recurrent fevers)
Familial medullary thyroid carcinoma (FMTC)
Familial paraganglioma and phaeochromocytoma
Familial parathyroid carcinoma
Familial periodic fever
Fanconi-Anämie, Typ C
Fascioscapulohumeral muscular dystrophy 1 (FSHD1)
Fascioscapulohumeral muscular dystrophy 2 (FSHD2)
Fatty liver, acute, of pregnancy
Fazio-Londe disease
Feingold syndrome 1
Fetal akinesia deformation sequence
Fragile X Associated tremor ataxia (FXTAS)
Fragiles X Syndrom
Friedreich'sche Ataxie (FRDA)
Frontotemporal dementia
Frontotemporal lobar degeneration, TARDBP-related
Frontotemporal lobar degeneration with ubiquitin-positive inclusions
Galactosemia 1, galactosaemia I, GALAC1
Gastrointestinal stromal tumor (GIST)
Gaucher disease, perinatal lethal
Gaucher disease, type I

Gaucher disease, type II
Gaucher disease, type III(A)
Gaucher disease, type IIIC
Gerstmann-Straussler disease
Glanzmann thrombastenia
Glaucoma
Glioma susceptibility 1 (GLM1), Glioblastoma
Glutaric acidemia
Glycogen storage disease of heart, lethal congenital
Glycogen storage disease, type 1A
Glycogen storage disease, various types
Gnathodiaphyseal dysplasia
Goodpasture syndrome
Haematuria
Halperin-Birk syndrome (HLBKS)
HARP syndrome
Heart-hand syndrome, Slovenian type
HELLP syndrome, maternal, of pregnancy
Hemolytic anemia, G6PD deficient (favism)
Hep. C virus (susceptibility, resistance, response to treatment)
Hereditary amyloidosis
Hereditary angiodema, type III
Hereditary angioedema 1
Hereditary deafness (DFNB1A)
Hereditary fructose intolerance
Hereditary Haemochromatosis Type 1 (HFE1)
Hereditary hypoceruloplasminemia
Hereditary motor and sensory neuropathy, type IIc
Hereditary nonpolyposis colorectal cancer syndrome (HNPCC), type 2
Hereditary nonpolyposis colorectal cancer syndrome (HNPCC), type 4 (Lynch syndrome 4)
Hereditary nonpolyposis colorectal cancer syndrome (HNPCC), type 5 (Lynch syndrome 5)
Hereditary nonpolyposis colorectal cancer syndrome (HNPCC), type 8 (Lynch syndrome 8)
Hereditary nonpolyposis colorectal cancer syndrome, type 1 (Lynch syndrome)
Hereditary spastic paraplegia
Huntington disease-like 1
Hutchinson-Gilford progeria
Hyperaldosteronism, familial, type II
Hyperalphalipoproteinema
Hyperbilirubinämie (M. Meulengracht; Gilbert's syndrome)
Hyperferritinemia-cataract syndrome
Hyperhomocysteinämie (MTHFR)
Hyper-IgE recurrent infection syndrome
Hyperkalemic periodic paralysis
Hyperlipoproteinämie
Hyperparathyroidism, familial, isolated
Hyperphenylalaninemia, BH4-deficient, B
Hypobetalipoproteinemia
Hypocalciuric hypercalcemia

Hypochondroplasia
Hypokalemic periodic paralysis
Ichthyosis, spastic quadriplegia, and impaired intellectual development
Immunodeficiency
Immunodeficiency-centromeric instability-facial anomalies syndrome
Inclusion body myopathy and brain white matter abnormalities (IBMWMA)
Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia
Infantile neuroaxonal dystrophy 1
Inherited cancer (sequencing panel)
Inherited metabolic disorders
Insomnia, fatal familial
Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies
Kagami-Ogata syndrome
King-Denborough syndrome
Klinefelter syndrome (47,XXY)
Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism
Kosaki overgrowth syndrome
Kufor-Rakeb syndrome
L-ferritin deficiency
Langer mesomelic dysplasia
Lateral meningocele syndrome
LCHAD deficiency
Leber congenital amaurosis
Leber hereditary optic neuropathy (LHON)
Legius syndrome
Left ventricular noncompaction 3
Leigh syndrome
Leri-Weill dyschondrosteosis
Lethal congenital contracture syndromes
Leukoencephalopathy with ataxia
Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation
Leukoencephalopathy with vanishing white matter 1/2/3/4/5, with or without ovarian failure
Lewy body dementia
Li Fraumeni syndrome
Limb Girdle muscular dystrophies (LGMD)
Lipodystrophy, congenital generalized, type 4
Lipodystrophy, familial partial, type 2
Lodder-Merla syndrome type 1 (LDMLS1)
Lodder-Merla syndrome type 2 (LDMLS2)
Long QT syndrome
Lungenkrebs, Bronchialkarzinom (NSCLC)
Machado-Joseph disease
Malignant hyperthermia susceptibility
Malouf syndrome
Mandibuloacral dysplasia
Marden-Walker syndrome
Marinesco-Sjogren syndrome
Marfan Syndrome (MFS)

Maturity Onset Diabetes of the Young (MODY) / Monogenic Diabetes
McArdle disease
Meningioma, SIS-related
Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration
Metachromatic leukodystrophy; MLD
Metachromatic leukodystrophy; MLD
Metatropic dysplasia
Microdeletion syndromes
Minicore myopathy with external ophthalmoplegia
Mismatch repair cancer syndrome 4 (MMRCS4)
Mitochondrial complex I deficiency, nuclear type 20
Mitochondrial complex III deficiency, nuclear type 2
Mitochondrial complex iv Deficiency, nuclear type 18
Mitochondrial cytopathies
Mitochondrial DNA deletion syndromes (keine näheren Angaben)
Mitochondrial DNA depletion syndrome 1 (MNGIE type)
Mitochondrial DNA depletion syndrome 12 (cardiomyopathic type)
Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)
Mitochondrial DNA depletion syndrome 16 (hepatic type)
Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type)
Mitochondrial DNA depletion syndrome 2 (myopathic type)
Mitochondrial DNA depletion syndrome 4A
Mitochondrial DNA depletion syndromes 4B
Mitochondrial myopathy and ataxia
Mitochondrial myopathy, encephalopathy, lactic acidosis and stroke-like episodes (MELAS)
Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy
Mitochondrial myopathy, isolated
Mitochondrial recessive ataxia syndrome
Mitochondrial trifunctional protein deficiency
Miyoshi muscular dystrophy
Morbus Crohn / Inflammatory Bowel Disease 1 (IBD1)
Morbus Waldenström / Macroglobulinemia Waldenstrom, Susceptibility to
Motor neuron disease
Mukolipidose, Typ IV
Multiple Endocrine Neoplasia Type 1
Multiple Endocrine Neoplasia Type 2
Multiple endokrine Neoplasie, Typ 2A (MEN2A)
Multiple endokrine Neoplasie, Typ 2B (MEN2B)
Multiple pterygium syndrome, lethal type
Muscle glycogenosis
Muscular dystrophy, congenital
Muscular dystrophy, congenital, due to ITGA7 deficiency
Muscular dystrophy, congenital, megaconial type
Muscular dystrophy, congenital, merosin deficient or partially deficient
Muscular dystrophy, congenital hearing loss, and ovarian insufficiency syndrome
Muscular dystrophy, congenital, with cataracts and intellectual disability
Muscular dystrophy, limb-girdle
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A

Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type B
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type C
Muscular dystrophy-dystroglycanopathy (limb-girdle)
MUTYH-associated polyposis
MYD88 deficiency (recurrent pyogenic bacterial infections)
Myasthenic syndrome, congenital, slow-channel
Myasthenic syndrome, congenital, fast-channel
Myasthenic syndrome, congenital
Myasthenic syndrome, congenital, with tubular aggregates
Myasthenic syndrome, congenital, without tubular aggregates
Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency
Myasthenic syndrome, congenital, presynaptic
Myasthenic syndrome, congenital, with pre- and postsynaptic defects
Myelofibrosis
Myeloproliferative disorder with eosinophilia
Myeloproliferative Neoplasms
Myoclonus epilepsy with ragged red fibres (MERRF)
Myoclonus, intractable, neonatal
Myofibromatosis, infantile
Myoglobinuria, acute recurrent, autosomal recessive
Myopathy, distal, 3
Myopathy, distal, 5
Myopathy, distal, 6, adult-onset, autosomal-dominant (MPD6)
Myopathy, distal, Tateyama type
Myopathy, distal, with anterior tibial onset
Myopathy, distal, with rimmed vacuoles
Myopathy, epilepsy, and progressive cerebral atrophy
Myopathy, myofibrillar
Myopathy, myofibrillar, fatal infantile hepyrtonic, alpha-B crystallin-related
Myopathy, scapulohumeroperoneal
Myopathy, tubular aggregate, 2
Myopathy with extrapyramidal signs
Myopathy with lactic acidosis, hereditary
Myopathy, X-linked, with excessive autophagy
Myosclerosis, congenital
Myositis (Idiopathic inflammatory myopathy, IM)
Myotonia congenita, atypical, acetazolamide-responsive
Myotonic Dystrophie Typ 1 (DM1)
Myotonic Dystrophie Typ 2 (DM2)
Narkolepsie
Nemaline myopathy, autosomal dominant
Nemaline myopathy, autosomal recessive
Neurodegeneration with ataxia, dystonia , and gaze palsy, childhood-onset
Neurodegeneration with brain iron accumulation
Neurofibromatosis Type 1
Neurofibromatosis Type 2
Neuromuscular disease, congenital, with uniform type 1 fiber
Neuronopathy, distal hereditary motor, autosomal dominant (HMND)

Neuronopathy, distal hereditary motor, autosomal recessive
Neuropathy, ataxia and retinitis pigmentosa (NARP)
Neuropathy, Hereditary Motor Sensory and Autonomic Type III
Neuropathy, hereditary sensory, type IE
Niemann-Pick disease
Nonaka myopathy
Non-Hodgkin Lymphoma (NHL)
Noonan syndrome
Oculopharyngeal muscular dystrophy 2
Omenn syndrome
Optic atrophy 12 (OPA12)
Osteogenesis imperfecta, type 1
Osteogenesis imperfecta, type 2
Osteogenesis imperfecta, type 3
Osteogenesis imperfecta, type 4
Osteoporosis
Ösophaguskarzinom
Ovarialkarzinom (genom. Instabilität)
Paget disease of bone 3
PAI1 Mangel (Hyperfibrinolyse)
Paramyotonia congenita
Parastremmatic dwarfism
Parkinsons Disease
Pearson marrow-pancreas syndrome
Pemphigus vulgaris, familiär (PV)
Peutz-Jeghers syndrome
Pfeiffer syndrome
Phenylketonuria
Phosphoglycerate kinase 1 deficiency
Pick disease
Platelet disorder, familial, with associated myeloid malignancy
Pneumothorax
POLG-related disorders
Polyglucosan body disease, adult form
Polyglucosan body myopathy 1/2
Polymicrogyria, bilateral temporooccipital
Polycystic kidney disease 4 with or without polycystic liver disease; PKD4; ARPKD
Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1/2
Pontocerebellar hypoplasia
Poor drug metabolism, Cyp2C19-related - Opremazol, Proguanil, Clopidogrel, Mephenytoin
Poor drug metabolism, Cyp2D6-related - Debrosoquin, Spartein, Notriptylin, Codein
Poor drug metabolism, Cyp3A4-related - Tacrolimus, Cyclosporin A
Porphyria, congenital erythropoietic (Gunther disease, CEP)
Porphyria cutanea tarda
Porphyria variegata
Porphyria, acute intermittent (AIP)
Prader-Willi-Syndrom (Angelmann-Syndrom)
Premature Ovarian Failure

Premature aging syndrome, Penttinen type
Primary lateral sclerosis, juvenile (PLSJ)
Progressive external ophthalmopegia, autosomal dominant 1
Progressive external ophthalmopegia, autosomal recessive 1
Progressive external ophthalmoplegia with mitochondrial DNA deletions
Prostate Cancer
Prothrombin Mangel, angeboren
Protoporphyria, erythropoietic, 1; EPP1
Pseudohypoparathyroidism type 1A (Albright hereditary osteodystrophy)
Pseudopseudohypoparathyroidism
Psoriasis vulgaris (Psoriasis 1 susceptibility)
Pulmonary Arterial Hypertension (PAH)
Pyogenic sterile arthritis, pyoderma gangrenosum, and acne
Renal tubular dysgenesis
Respiratory insufficiency
Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities
Retinitis pigmentosa
Retinoblastoma
Retinopathies (Retinitis Pigmentosa, Cone/Cone-rod dystrophies, Leber congenital amaurosis and macular degeneration
Rett-Syndrom
Rhabdomyosarcoma 2, alveolar
Rheumatoide Arthritis (RA)
Rippling muscle disease 2 Rippling muscle disease 2
Russell-Silver syndrome
Sandhoff disease
Sarcoma
Scapuloperoneal spinal muscular atrophy
Scapuloperoneal syndrome, neurogenic, Kaeser type
Schwannomatosis
Sclerosteosis 2
Seckel syndrome
SED, Maroteaux type
Severe combined immunodeficiency, Athabascan type
Severe combined immunodeficiency, B cell-negative
Severe combined immunodeficiency due to ADA deficiency
Severe Combined Immunodeficiency (pilot)
Short stature, idiopathic familial / Familial SHOX-related disorders
Sialuria
Sichelzellanämie
Silver-Russell Syndrom (SRS)
single mtDNA deletion disorders
Sneddon syndrome
Spastic ataxia 1
Spastic ataxia 5 (SPAX5)
Spastic paralysis, infantile-onset ascending; IAHSP
Spinal and bulbar muscular atrophy, X-linked 1 (SMAX1; Kennedy disease)
Spinal muscular atrophy, infantile, James type (SMAJI)
Spinal muscular atrophy, Jokela type

Spinal muscular atrophy, late-onset, Finkel type
Spinal muscular atrophy, lower extremity-predominant, autosomal dominant
Spinal muscular atrophy, type 1
Spinal muscular atrophy, type 2
Spinal muscular atrophy, type 3
Spinal muscular atrophy, type 4
Spinal muscular atrophy with congenital bone fractures
Spinal muscular atrophy with progressive myoclonic epilepsy
Spinocerebellar ataxia
Spinocerebellar ataxia 1
Spinocerebellar ataxia 2
Spinocerebellar ataxia 6
Spinocerebellar ataxia 7
Spinocerebellar ataxia, autosomal recessive 8
Spinocerebellar ataxia type 3
Spinocerebellar ataxia type 4
Spinocerebellar ataxia type 5
Split-foot malformation with mesoaxial polydactyly
Spondyloarthropathy 1 (Spondylitis ankylosans, M. Bechterew)
Spondylometaphyseal dysplasia, Kozlowski type
Spongiform encephalopathy with neuropsychiatric features
Stargardt disease 3
Stevens-Johnson Syndrome, Allopurinol induced (ASJ)
Stevens-Johnson Syndrome, Carbamezepine induced (CSJ)
Stickler syndrome type I
Stickler Syndrome Type II
Stickler syndrome type IV
Stickler syndrome type V
Stormorken syndrome
Structural eye abnormalities: Microphtalmia/anophtalmia, Coloboma, Anterior chamber defects
Systemic hemosiderosis due to aceruloplasminemia
Tay Sachs disease (TSD)
Temple syndrome (UPD14)
Thalassämie alpha
Thalassämie beta
Thanatophoric Dysplasia
Thiopurin-S-Methyltransferase (TPMT) Mangel
Thrombocytemia 1
Thrombophilia due to protein C deficiency, AD (THPH3)
Thrombophilia due to protein C deficiency, AR (THPH4)
Thrombophilia due to protein S deficiency, AD (THPH5)
Thrombophilia due to protein S deficiency, AR (THPH5)
Thrombophilie (activated protein C resistance; THPH2)
Thrombophilie (THPH1, F5, F2, (MTHFR))
Thyrotoxic periodic paralysis, susceptibility to, 1
Tibial muscular dystrophy, tardive
Triploidy
Trisomy 13 (Patau Syndrome)

Trisomy 18 (Edwards syndrome)
Trisomy 21 (Down Syndrome)
Tuberus sclerosis complex (TSC)
Turner Syndrome
Ullrich congenital muscular dystrophy 1
Ullrich congenital muscular dystrophy 2
Urothelial cell carcinoma
Uruguay faciocardiomusculoskeletal syndrome FCMSU
Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome
VEXAS syndrome, somatic
Vici syndrome
Vitamin D-dependent rickets, type 2A
VLCAD deficiency
Von Hippel-Lindau syndrome
Welander distal myopathy
Williams-Beuren syndrome
Wilson disease
Wolff-Parkinson-White syndrome (WPW syndrome)
X inactivation
Y-chromosome microdeletions
Y deletion
Yunis-Varon syndrome
Zervixkarzinom

	Anbieter & Methoden							
OMIM	BVDH	CAP	CF Network	EMQN	EQUALIS	ESP-EQA	ETRL	GenQA
#250950				NGS, CNV				
		S						
#201450		S						X
								X
#100800				S				X
#613737				NGS, CNV				
#613065								FISH, X
#601626					S			FISH, X
#102700				NGS, S				
#300100				NGS, CNV				
#300100				NGS, CNV				
#300755								X
								X
#203450				NGS, CNV				
#613490		S						
#609889				NGS, S				
#301050								X
#614820				NGS, CNV				
#104310		S			S			X
# 580000								X
#105400				S, NGS, CNV				X
#606353				NGS, CNV				
#170390				NGS, S				
#300068								X
								X
#105830		S, MET		MET, SB				X
#613118								
#101200								X
#607485				NGS, CNV				
				S, NGS				
#193700				S, NGS				
#618436				S, NGS				
#617146				S, NGS				
#618484				S, NGS				
#619334				S, NGS				
# 208900								S
#604391				S				
				NGS, CNV				X
#615952								
#173900				S, NGS, CNV				S
#617383				NGS, S				
#615988				NGS, S				
				NGS, CNV				
#300376		S		S, CNV				X
#130650				S, CNV, MET				X
#158810				NGS, S				

#616471				NGS, S				
#613812				NGS, CNV				
#605808		S						
#135150								
#186580				S, NGS, CNV				
#187800								
# 619271								
#614200								
#210900		S						X
#603457				S, MET, CNV				
#113500				NGS, S				
#604370		S, FISH		S, NGS, CNV				X
								X
				NGS, S				
#611777								X
#617936								
#271900		S						
#601338				NGS, CNV				
#600142				NGS, CNV				
#608579		S						
				S, NGS, CNV				X
		S		S, NGS, CNV				X
#607487				NGS, S				
#254940				NGS, S				
#212140				NGS, S				
#115430				NGS, S				
#613763				NGS, S				
				X				X
#212780				NGS, S				
#117000				S, NGS, CNV				
#209880				S, NGS				
				S, NGS				
#604290				NGS, CNV				
#212840				NGS, CNV				
#604121				NGS, CNV				
#137440				NGS, CNV				
				NGS, CNV				
#213700				NGS, CNV				
#614706				NGS, CNV				
#615362				NGS, CNV				
#162350				NGS, CNV				
#601780				NGS, CNV				
#204300				NGS, CNV				
				S, NGS, CNV				X
#214500				NGS, CNV				
								X
#143100		S		S				X

#200150				NGS, CNV				
								X
#123320				NGS, S				
								X
#151400								FISH, X
#608232								FISH, X
#212750		S						
#114500				S, NGS		X		X
#233650				NGS, S				
#615182				NGS, S				
#607473				S, NGS, CNV				
#610505				NGS, S				
#300985								X
#201910				S, NGS, SB, CNV				X
#202400								
				NGS, S				
#616004								
# 618494								X
#223000					S			
								X
				NGS, S, CNV				
#620246				NGS, S				
#602771				NGS, S				
#605637				NGS, S				
				NGS, S				
								S
#614563				NGS, S				
#122700								
								X
				NGS, S				
#123400				NGS, CNV				
#123500								X
#154800				S, NGS				
#219700		S	X	S				X
#300257				S, NGS				
		S						
				NGS, CNV				
# 125370								X
#607907				NGS, CNV				
				NGS, CNV				
#613217				S, NGS, CNV				
#188400								X
#606835				NGS, S				
#274270				S, NGS, CNV				
#607208								X
#310200		S		S, CNV				X
#128235				NGS, CNV				
#616411				NGS,S				

#128230				NGS, CNV				
#145680				S, NGS				
								X
				S, NGS				X
#617900				NGS, CNV				
#614212				S, NGS				
# 608089								X
				NGS, S				
#254780				NGS, CNV				
#254780				NGS, CNV				
				S, CNV				
#265000				S, CNV				
#615363				S, CNV				
#301500								X
#228000				NGS, S				
#227500								
#234000								
#613225								
#227400		S			S			
#175100				S, NGS, CNV				X
#613980				NGS, S				
								X
#143890				S				X
#144010				S				X
#603776				S				X
								X
#249100				S				
#155240				S, NGS, CNV				X
#168000								X
#145000								X
#142680				S, NGS, CNV				
#227645		S						X
#158900				S				
#158901				S, MET, CNV				
#609016				S, NGS				
#211500				NGS, S				
#164280		NGS						
				S, NGS				
		S						X
#300624		S		S, MET, SB, X				X
#229300		S		S, SB				X
#600274				NGS, CNV				X
#612069				NGS, CNV				
#607485				NGS, CNV				
# 230400								X
#606764				S, NGS				X
#608013		S						
#230800		S						

#230900		S						
#231000		S						
#231005		S						
#137440				NGS, CNV				
#273800								
				NGS, CNV				
#137800		S, NGS						X
				NGS, S				
#602743				NGS, S				
#232200		S						X
				NGS, S				
#166260				NGS, S				
#233450								
#141200								X
#618651				S				
#607236				NGS, CNV				
#610140				NGS, S				
#609016				NGS, S				
#300908		S						
#609532								
#105210				NGS, S				
#610618								
# 106100								X
#220290		S		S, NGS, CNV				
#229600								
#235200		S		S, NGS	S			
#604290				NGS, CNV				
#606071				NGS, CNV				
#609310		X		S, NGS, CNV				X
#614337		X		S, NGS, CNV				X
#614350		X		S, NGS, CNV				X
#613244				S, NGS, CNV				
#120435		X		S, NGS, CNV				X
				NGS, CNV				X
#603218				NGS, CNV				
#176670				S, NGS				
#605635				NGS, CNV				
#143470								
#143500		S		S, NGS, CNV				X
#600886				NGS, CNV				
#236250		S			S			
#147060								
#170500				NGS, S				
#617347		S						
# 145000								
#233910				NGS, CNV				
#615558				S, NGS, CNV				

#146000				S				X
				NGS, S				
#614457				NGS, CNV				
		FISH		NGS, S				
								S
#619733				NGS, CNV				
				NGS, CNV				
#256600				NGS, CNV				
		S						
		X						
#600072				NGS, CNV				
#619031				NGS, S				
# 608149								X
#145600				S, NGS, CNV				
								X
#616549				NGS, S				
#616592				NGS, CNV				
#610513				NGS, CNV				
#615604				NGS, CNV				
#249700				S, NGS, CNV				
#13720				NGS, CNV				
#609016				S, NGS				
								X
#535000		S		S, NGS				X
# 611431								X
#601493				NGS, S				
#256000				S, CNV				X
#127300				S, NGS, CNV				
				NGS, S				
#615651				NGS, CNV				
#611105				NGS, CNV				
				NGS, CNV				
#127750				NGS, CNV				
# 151623								X
#608099				NGS, S				X
#613327				NGS, S				
#151660				NGS, S				
#617173				NGS, S				
#617182				NGS, S				
				X, NGS, S				X
#211980		IHC		X		X, FISH		X
#109150				S				
				S, NGS, CNV				
#212112				NGS, S				
#248370				NGS, S				
#248700				NGS, S				
#248800				NGS, S				
#154700								X

#606391								
#232600				NGS, S				
#607174								
#616878				NGS, S				
#250100				NGS, CNV				
#250100				NGS, CNV				
#156530				NGS, S				
								X
#255320				S, NGS, CNV				
# 619101								
#611126				NGS, CNV				
#615157				NGS, CNV				
#619062				NGS, S				
		S						X
		S						
#603041				NGS, S				
#612075				NGS, S				
#612073								
#618528				NGS, S				
#619425				NGS, S				
#609560				NGS, S				
#203700								
#613662								
#617675				NGS, S				
#540000				S, NGS				
#251900				S, NGS				
#616209				NGS, CNV				
#607459								
				NGS, S				
				NGS, S				
#266600				S, NGS, CNV				
#153600								
#252650		S						
#131100								X
#171400		S		S, NGS, CNV				X
#171400		S		S, NGS, CNV				X
#162300		S		S, NGS, CNV				
#253290				NGS, S				
#300559				NGS, S				
#613205				NGS, S				
#613204				NGS, S				
#602541				NGS, S				
#607855				NGS, S				
#619518				NGS, S				
#617404				NGS, S				
				NGS, S				
				NGS, S				

				NGS, S				
				NGS, S				
				NGS, S				
#608456				S, NGS, CNV				X
#612260								
				NGS, S				
				NGS, S				
				NGS, S				
				NGS, S				
				NGS, S				
				NGS, S				
#254210				NGS, S				
#615120				NGS, S				
#254450					S			
#131440				NGS, CNV				
#616871								FISH, NGS, X
#545000		S		S, NGS				NGS, X
#617235				NGS, CNV				
				NGS, CNV				
#268200				S, NGS				
#610099				NGS, CNV				
#617030				NGS, CNV				
#618655				S, NGS				
#614321				S, NGS				
#606768				S, NGS				
#617158				NGS, CNV				
#619036				NGS, S				
				NGS, S				
#613869				NGS, S				
#616852				NGS, CNV				
#615883				NGS, S				
# 615673				NGS, S				
#255125				NGS, S				
#310440								
#255600				NGS, S				
#160750								
#608390				NGS, S				
#160900		S		S, SB				X
#602668				S, SB				
#161400		S						
				NGS, S				
				NGS, S				
#617158				NGS, CNV				
				NGS, CNV				
#162200								X
#101000								X
#117000				S, NGS, CNV				
				S, NGS				

				S, NGS				
#551500				S, NGS				X
#223900		S						
#614116				NGS, CNV				
#257200		S						
#605820				NGS, S				
#605027								NGS, FISH, X
#163950								X
#620460				NGS, S, CNV				
#603554				NGS, S				
#618977				NGS, CNV				
#166200				S, NGS, CNV				X
#166210				S, NGS, CNV				X
#259420				S, NGS, CNV				X
#166220				S, NGS, CNV				X
#166710								
#167250				NGS, CNV				
#613329		S						
#168300				NGS, S				
#168400				NGS, S				
#168600				NGS, CNV				
#557000				S, NGS				
#169610		S						
#175200								X
#101600								X
#261600				S				
#300653				NGS, S				
#172700				NGS, CNV				
# 601399								
# 173600								X
								X
#263570				NGS, S				
				NGS, S				
#612691				NGS, CNV				
# 263200								X
#221770 / #618193				NGS, CNV				
				NGS, CNV				
#609535		S						
#608902		S						
		S						
#263700				S, NGS, CNV				
#176100				S, NGS, CNV				
#176200				S, CNV				
#176000				S, NGS, CNV				
#176270		S, MET		MET, SB				FISH, X
								X

#601812				NGS, CNV				
#606353				NGS, CNV				
#157640								
#258450								
				NGS, S				
		S		S				X
#613679		S						
#177000				S				
# 103580								X
# 612463								X
#177900		S						
#178600								
#604416				S				
# 267430								
								X
***603904**				NGS, CNV				
				S, NGS				X
#180200				S, NGS, CNV				
				S, NGS				X
#312750		S						X
#268220				NGS, S				
#604302								
#606072				NGS, S				
#312780								X
#268800				NGS, CNV				
		S, X						X
#181405				NGS, S				
#181400				NGS, S				
								X
#614305				NGS, S				
								S
#184095				NGS, S				
#602450				NGS, S				
#601457				NGS, S				
#102700				NGS, S				
#601457								X
#300582				S, NGS, CNV				
#269921				NGS, S				
#603903								
#180860				S, CNV, MET				X
								X
#182410				S				
#108600				NGS, CNV				
#614487				NGS, CNV				
#607225				NGS, CNV				
#313200								X
#619042				S, NGS				
#615048				NGS, CNV				

#182980				NGS, CNV				
				S, NGS				
#253300		S, CNV		S				X
#253550		S, CNV		S				
#253400		S, CNV		S				
#271150		S, CNV		S				
#616866				NGS, S				
#159950				NGS, S				
				NGS, CNV				X
#164400		S		S				
#183090		S		S				
#183086				S				
#164500				S				
#610743				NGS, S				
#109150		S		S				
#600223		S		S				
#600224		S		S				
#616890				NGS, S				
#106300		S						
#184252				NGS, S				
#606688				NGS, CNV				
#600110				NGS, CNV				
#608579								
#108300				S, NGS, CNV				
#604841				S, NGS, CNV				
#614134				S, NGS, CNV				
#614284				S, NGS, CNV				
#185070				NGS, S				
								X
#604290				NGS, CNV				
#272800		S		NGS, CNV				
# 616222								X
#301040		S						
#603902		S						
#187600				S				X
#610460		S		S, NGS, CNV				X
#187950								
#176860								
#612304								
#612336								
#614514								
#188055								
#188050		S			S			
#188580				NGS, S				
#600334				NGS, S				
								X
#264480								NGS, FISH, X

#601161								NGS, FISH, X
#190685								NGS, FISH, X
#191100								X
#309590								NGS, FISH, X
#254090				NGS, S				
#616470				NGS, S				
		FISH		S				
#300280				S, NGS				
#615688				S				
#301054				S, NGS				
#242840				S, NGS				
#277440								
#201475				NGS, CNV				
#193300				S, NGS, CNV				X
#604454				NGS, S				
#194050								X
#277900				S, NGS, CNV				
#194200				S, NGS				
								X
#415000				X				
								X
#216340				NGS, CNV				

INSTAND	ÖQUASTA	QuIP	RCPA QAP	RfB (DGKL)	UHKT
NGS			S, NGS		
S				S	
S				S	
S				S	
S				S	
S					
S					

				S	
		IHC			
				S	
S					
				S	
S		S		S	
				S	
		S			
S				S	

			FISH		
S			S, X		S
		S	S		
S				S	
S				S	
				S	
				S	
S				S	
S				S	
S				S	
S				S	
S					
S					

S					
				S	
				S	
				S	
S				S	
S				S	
S					
S				S	
		S, IHC		S	

S				S	
			S, MET, X		
S					
				S	
				S	
S				S	
S			S	S	
S				S	
S				S	
S				S	
S				S	
				S	
S				S	
S				S	
S					
				S	
S					
S				S	
S					

		S, IHC	S, FISH, IHC		

S					
S					
				S	
S					
S					
S					
S					

S					
S			S		
			IHC		
					S

				S	
		IHC			
		S			
S				S	
NGS					
S					
S					
S				S	

	IHC, S				
S					
S					
S					
				S	
		S, IHC, FISH			
			S		

S				S	
S				S	
S				S	
S				S	
				S	S
			S		
			S		
S					
				S	
			S		

				S	
S				S	
S				S	
		IHC			

Methode		
	BVDH	CAP
Array-Diagnostik	X	
Cell free DNA detection (common microdeletions; noninvasive prenatal testing)		
Cell free DNA detection (fetal aneuploidy; noninvasive prenatal testing)		X
Cell free DNA detection (fetal sexing; noninvasive prenatal testing)		
Cell free DNA detection (oncogenic mutations)		X
Constitutional CNV - postnatal		
Constitutional CNV - pränatal		
Cytogenomic microarray		X
DNA extraction (Blood)		X
DNA extraction (Fresh frozen tissue)		
DNA extraction (Saliva)		
DNA extraction and amplification from formalin-fixed, paraffin-embedded tissue		X
DNA quantification		
DNA sequencing		X
FISH (fixierte Zellen)	X	X
FISH (Paraffin-embedded tissues)		X
Genetische Beratung	X	
IHC (Immunohistochemistry)		X
Molecular tissue identification (Molecular fingerprinting)		
NGS (Next generation sequencing)	X	X
PID/PGT: Blastomere testing by FISH (aneuploidy, strucural rearrangements)		
PID/PGT: Polar bodies testing by NGS and/or arrays		
PID/PGT: Trophectoderm/blastomere testing for aneuploidies		
PID/PGT: Trophectoderm/blastomere testing for chromosomal rearrangements		
PID/PGT: Trophectoderm/blastomere testing for monogenetic disorders		
Whole exome sequencing		
Zytogenetik (Chromosomenaberrationen)		X
Zytogenetik (Karyotypisierung, Chromosomenbandanalyse)	X	X
Zytogenetik postnatal	X	
Zytogenetik pränatal: Chorionzottenbiopsie (CVS)	X	
Zytogenetik pränatal: Fruchtwasseranalyse (Karyotypisierung)	X	
Zytogenetik pränatal: Schnelltest (FISH, PCR)	X	
Constitutional Cytogenetics - Postnatal		
Cytogenetics	X	X
Molecular Rapid Aneuploidy (MRA): QF-PCR, MLPA, BoBs		
Chromosome Breakage Syndromes		
Zytogenetik, molekular	X	

Glycogen storage disease, type 1A

	Anbieter								
CF Network	EMQN	EQUALIS	ESP-EQA	ETRL	GenQA	INSTAND	ÖQUASTA	QuIP	RCPA QAP
					X				
	X				X				
	X				X				
	X				X				
					X				
					X				
					X				X
					X				
					X				X
					X				
	X	X							X
									X
					X				
									X
					X				
	X				X			X	
					X				
					X				
					X				
					X				
	X				X				
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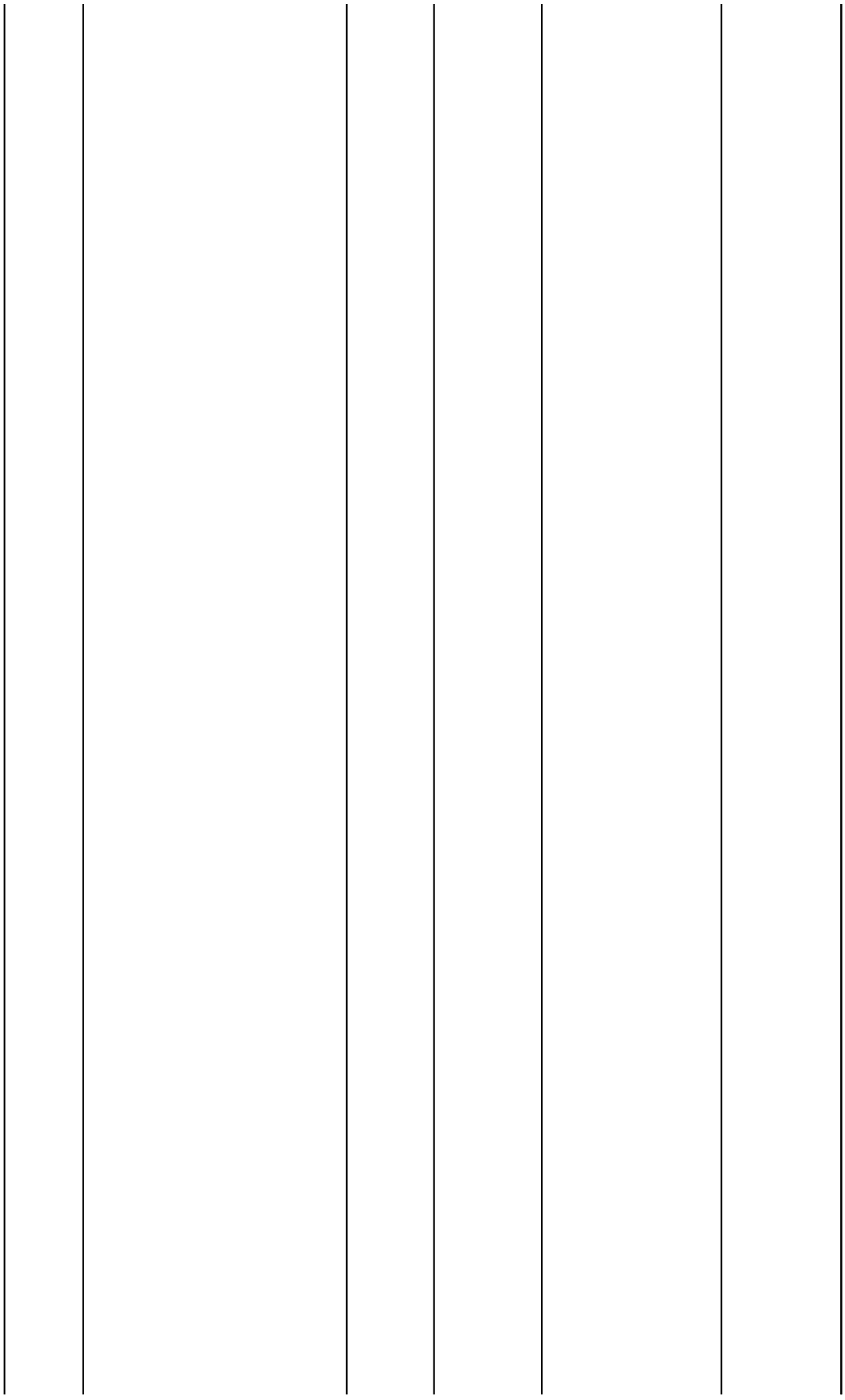
					X				
					X				
					X				
					X				
					X				



RfB (DGKL)	UHKT
X	
X	
X	