Diagnostic offer **Gene panels**

SYNLAB MVZ
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Opthalmology

☐ **Achromatopsia [GP001] (~10 kb)**
ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H

☐ **Albinism, ocular [GP002] (~21 kb)**
1. GPR143; 2. C10orf11, LYST, MC1R, OCA2, SLC45A2, TYR, TYRP1

☐ **comprehensive diagnostic [GP002XL]** (69,5kb)
AP3B1, BLOC1S3, BLOC1S6, C10orf11, DTNBP1, EPG5, EDN3, EDNRB, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, MC1R, MITFSNAI2, MLPH, MYO5A, OCA2, PAX3, RAB27A, SLC24A5, SLC45A2, SOX10, TYR, TYRP1

☐ **Albinismus, oculocutaneous [GP168] (~10 kb)**
MC1R, OCA2, SLC24A5, SLC45A2 (MATP), TYR, TYRP1

☐ **Axenfeld-Rieger syndrome [GP172]**
ASPH, B3BLCT, COL4A1, COL4A2, CYP1B1, EYA1, FOXC1, FOXE3, HMX1, LTBP2, MIR184, MYOC, PAX6, PITX2, PXDN, SLC38A8

☐ **Hermansky-Pudlak syndrome [GP167] (~22 kb)**
AP3B1, AP3D1, BLOC1S3, BLOC1S6, DTNBP1, HPS1 HPS3, HPS4, HPS5, HPS6, PLDN

☐ **Bardet-Biedl syndrome [GP003] (~19 kb)**
BBS1, BBS10, BBS12, BBS2, BBS4, BBS7, BBS9, MKKS, MKS1, TTC8

☐ **comprehensive diagnostic [GP003XL]**
ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CCDC28B, CEP290, LZTFL1, MKKS, MKS1, NPHP1, SDCCAG8, TMEM67, TRIM32, TTC21B, TTC8, WDPCP

☐ **Joubert syndrome [GP004] (~25 kb)**
AHI1, CC2D2A, CEP290, NPHP RPGRIP1L, TMEM67

☐ **comprehensive diagnostic [GP004XL]**
AHI1, ARL13B, B9D1, C5orf42, CC2D2A, CEP290, CEP41, CSPP1, KIF7, MKS1, NPHP1, OFD1, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM237, TMEM67, TTC21B

☐ **Liver hereditary optic neuropathy (LHON) [GP165]**
MT-ATP6, MT-CO1, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT - ND4, MT-ND4L MT-ND5, MT-ND6, MT-RNR1, MT-TE, MT-TL1, MT-TM, MT - TQ, MT- TT

☐ **Catarakt, congenital [GP005]** (~23 kb)
BFSP1, BFSP2, CRYAA, CRYAB, CRYBA1, CRYBB1, CRYBB2, CRYBB3, CRYGC, CRYGD, CRYGS, EPHA2, FYCO1, GJA3, GJA8, HSF4, LIM2, MAF, MIP

☐ **Leber congenital amaurosis (LCA) [GP006] (~23 kb)**
AIPL1, CEP290, CRX, GUCY2D, LCA5, RDH12, RPE65, RPGRIP1

☐ **comprehensive diagnostic [GP006XL]**
ABCA4, AHI1, AIPL1, ALMS1, CABP4, CEP290, CRB1, CRX, GDF6, GUCY2D, IFT140, IMPDH1, IQCB1, KCNJ13, KCNV2, LCA5, LRAT, MERTK, MPDZ, NMNAT1, NPHP1, NXNL1, OTX2, PRPH2, RD3, RDH12, RDH5, RPE65, RPGRIP1, RPGRIP1L, SPATA7, TULP1

☐ **Anophthalmia/microphthalmia/coloboma (MAC-spectrum) [GP007] (~25 kb)**
BCOR, BMP4, CHD7, FOXE3, GDF6, OTX2, PAX6, RAX, SOX2, STRA6

☐ **comprehensive diagnostic [GP007XL]**
BCOR, BMP4, CHD7, FOXE3, FREM1, GDF3, GDF6, HCCS, MFRP, MITF, NAA10, OTX2, PAX6, PRSS56, RAX, SHH, SIX6, SMOC1, SOX2, STRA6, VAX1, VSX2

☐ **Optic atrophy [GP009] (~19 kb)**
1. OPA1, 2. CISD2, MFN2, NDUFS1, NR2F1, OPA3, POLG, RTN4IP1, SPG7, TIMM8A, TMEM126A, WFS1

☐ **Peters Anomalie [GP171] (~6kb)**
CYP1B1, FOXC1, PAX6, PITX2, PITX3

☐ **Refsum-Syndrom [GP010] (7,7kb)**
PEX1, PEX2, PEX26, PEX7, PHYH

☐ **Retinitis pigmentosa [GP011]**
AD/XL: *IMPDH1, KLHL7, NR2E3, PRPF31, PRPF8, PRPH2, RHO, RP1, RP2, RPGR* (~ 25 kb)
AR: *EYS, USH2A* (~ 24 kb)

☐ **comprehensive diagnostic [GP011XL]**
ABCA4, AIPL1, ARL6, BEST1, C2orf71, C8orf37, CA4, CDHR1, CERKL, CLRN1, CNGA1, CNGB1, CRB1, CRX, DHDDS, EYS, FAM161A, FLVCR1, FSCN2, GUCA1B, IDH3B, IMPDH1, IMPG2, KLHL7, LRAT, MAK, MERTK, NR2E3, NRL, PDE6A, PDE6B, PDE6G, PRCD, PROM1, PRPF3, PRPF31, PRPF6, PRPF8, PRPH2, RBP3, RDH12, RGR, RHO, RLBP1, ROM1, RP1, RP1L, RP2, RP9, RPE65, RPGR, SAG, SEMA4A, SLC7A14, SNRNP200, SPATA7, TOPORS, TTC8, TULP1, USH2A, ZNF513

☐ **Stargardt disease/ macular Dystrophia [GP008] (~24 kb)**
1. ABCA4, 2. BEST1, CDH3, CNGB3, CRB1, ELOVL4, PROM1, PRPH2, RDH12, RP1L1, TIMP3

☐ **Stickler syndrome [GP013] (~22 kb)**
COL11A1, COL2A1, COL9A2, COL9A3, COL11A2, COL9A1

☐ **USHER syndrome [GP091] (~24 kb)**
1. USH2A, 2. MYO7A, USH1C

☐ **comprehensive diagnostic [GP091XL]**
CDH23, CIB2, CLRN1, DFNB31, HARS, MYO7A, PCDH15, PDZD7, USH1C, USH1G, USH2A

☐ **Cone-/cone-rod dystrophies (ZD/ZSD) [GP014] (~25 kb)**
ABCA4, ADAM9, CERKL, CNGA3, KCNV2, PDE6C, RPGRIP1, RDH5

☐ **comprehensive diagnostic [GP014XL]**
ABCA4, ACBD5, ADAM9, AIPL1, ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8orf37, CABP4, CACNA1F, CACNA2D4, CCDC28B, CDHR1, CEP290, CERKL, CNGA3, CNGB3, CNNM4, CRB1, CRX, CYP4V2, GNAT2, GUCA1A, GUCY2D, KCNV2, LZTFL1, MKKS, MKS1, NPHP1, PDE6C, PDE6H, PITPNM3, PROM1, PRPH2, RAB28, RAX2, RDH5, RGS9, RGS9BP, RIMS1, RPGR, RPGRIP1, SDCCAG8, SEMA4, TMEM67, TRIM32, TTC21B, TTC8, WDPCP

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Epilepsy and migraine

☐ **Dravet syndrome [GP016] (~21 kb)**

1. SCN1A, 2. GABRG2, SCN2A, SCN9A, STXBP1

☐ **Epilepsy [GP017] (~25 kb)**

1. SCN1A, 2. CDKL5, GABRA1, GABRG2, KCNQ2, PCDH19, STXBP1, SYNGAP1

□ **comprehensive diagnostic [GP017XL]**

ACY1, ADSL, ALDH7A1, AMT, ARHGEF9, ARX, CACNA1H, CACNB4, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN2, CNTNAP2, CPA6, CPT2, EFHC1, EPM2A, FOLR1, FOXG1, GABRA1, GABRB3, GABRD, GABRG2, GAMT, GCSH, GLDC, GRIN2A, GRIN2B, JRK, KCNJ10, KCNMA1, KCNQ2, KCNQ3, KCNT1, LGI1, MAGI2, MAPK10, MECP2, MEF2C, MTHFR, NRXN1, PCDH19, PLCB1, PNKP, PNPO, PRRT2, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SLC19A3, SLC25A22, SLC2A1, SLC9A6, SPTAN1, SRPX2, STXBP1, SYNGAP1, TBCE, TCF4, TREX1, UBE3A, ZEB2

☐ **Epilepsy, childhood absence (ECA) [GP015] (~14 kb)**

CACNA1H, GABRA1, GABRB3, GABRG2, JRK, SLC2A1

☐ **Epileptic encephalopathy, early infantile (EIEE; Ohtahara syndrome) [GP019XL]**

ARHGEF9, ARX, CDKL5, KCNQ2, PCDH19, PLCB1, PNKP, SCN1A, SCN2A, SCN8A, SLC25A22, SPTAN1, STXBP1

☐ **Epileptic encephalopathy [GP018] (~24kb)**

CDKL5, GABRA1, SCN1A, SCN2A, SCN8A, STXBP1

□ **comprehensive diagnostic [GP018XL]**

ACY1, ADAR, ADSL, ALDH7A1, ALG13, AMT, ARHGEF9, ARX, BRAT1, CACNA1A, CASK, CDKL5, CHD2, CNTNAP2, CPT2, DCX, DNM1, FLNA, FOLR1, FOXG1, GABRA1, GABRB3, GABRG2, GAMT, GCSH, GLDC, GPHN, GRIN1, GRIN2A, GRIN2B, HDAC4, HNRNPU, KCNJ10, KCNQ2, MAGI2, MAPK10, MBD5, MECP2, MEF2C, MOCS1, MOCS2, MTHFR, NRXN1, PCDH19, PLCB1, PNKP, PNPO, PRRT2, RNASEH2A, RNASEH2B, RNASEH2C, ROGDI, SAMHD1, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SLC19A3, SLC25A22, SLC2A1, SLC9A6, SPTAN1, ST3GAL3, ST3GAL5, STXBP1, SYN1, SYNGAP1, TBC1D24, TBCE, TCF4, TREX1, TSC1, TSC2, UBE3A, WDR45, ZEB2

☐ **Generalized epilepsy with febrile seizures plus (GEFS+) [GP020] (~21kb)**

1. SCN1A 2. GABRD, GABRG2, SCN1B, SCN2A, SCN9A

☐ **MELAS syndrome [GP164]**

MT-TL1, MT-ND1, MT-ND5, MT-ND4, MT-ND6, MT-CYB, MT-CO3, MT-CO1, MT-CO2, MT-ND3, MT-TF, MT-TQ, MT-TH, MT-TV, MT-TW, MT-TK, MT-TL2, MT-TE, MT-TC, MT-TS1, MT-TS2

☐ **Hyperekplexia, hereditary [GP021] (~9 kb)**

ARHGEF9, GLRA1, GLRB, GPHN, SLC6A5

☐ **Metabolic epilepsy [GP022XL]**

ACY1, ADSL, AGA, ALDH4A1, ALDH5A1, ALDH7A1, AMT, ARG1, ATIC, BTD, DPYD, ETFA, ETFB, ETFDH, FH, FOLR1, GAMT, GCDH, GCH1, GCSH, GLDC, GNE, GPHN, HPD, L2HGDH, MOCS1, MOCS2, MTHFR, PCBD1, PGK1, PNPO, PRODH, PTS, QDPR, SLC25A15, SLC46A1, SUOX

☐ **Autosomal Dominant Nocturnal Frontal Lobe Epilepsy (ADNFLE) [GP177] (~14kb)**

CRH, CHRNA2, CHRNA4, CHRNB2, DEPDC5, KCNT1

☐ **Familial hemiplegic migraine (FHM) [GP023] (~23 kb)**

ATP1A2, ATP1A3, CACNA1A, SCN1A

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Intellectual disability and Developmental delay

☐ **Bardet-Biedl syndrome [GP003] (~ 22 kb)**

ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, MKKS, MKS1, TTC8

☐ **comprehensive diagnostic [GP003XL]**

ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CCDC28B, CEP290, LZTFL1, MKKS, MKS1, NPHP1, SDCCAG8, TMEM67, TRIM32, TTC21B, TTC8, WDPCP

☐ **Cornelia-de-Lange syndrome (CdLS) [GP078] (~19 kb)**

1. NIPBL, 2. SMC1A, SMC3, RAD21, HDAC8

☐ **Dravet syndrome [GP016] (~21 kb)**

1. SCN1A, 2. GABRG2, SCN2A, SCN9A, STXBP1

☐ **Glycosylation disorders (CDG) [GP079] (~22 kb)**

1. PMM2, 2. MPI, ALG6, 3. ALG1, ALG12, ALG2, ALG8, ALG9, DOLK, DPAGT1, DPM1, MAGT1, MPDU1, RFT1 SRD5A3, TMEM165, TUSC3

☐ **Joubert syndrome [GP004] (~25 kb)**

AHI1, CC2D2A, CEP290, NPHP RPGRIP1L, TMEM67

☐ **comprehensive diagnostic [GP004XL]**

AHI1, ARL13B, B9D1, C5orf42, CC2D2A, CEP290, CEP41, CSPP1, KIF7, MKS1, NPHP1, OFD1, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM237, TMEM67, TTC21B

☐ **Zellweger syndrome [GP035] (~18 kb)**

1. PEX1, 2. PEX6, 3. ABCD3, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5

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Cortical malformations

☐ **Lissencephaly, type I [GP029] (~16kb)**

ARX, DCX, PAFAH1B1,RELN, TUBA1A

□ **comprehensive diagnostic see [GP032XL]**

☐ **Mikrocephaly, primary [GP031] (~24kb)**

ASPM (MCPH5), CDK5RAP2 (MCPH3), MCPH1 (MCPH1), STIL (MCPH7), WDR62 (MCPH2)

□ **comprehensive diagnostic see [GP032XL]**

☐ **Warburg-Mikro syndrome [GP034] (~9 kb)**

RAB3GAP1, RAB3GAP2, RAB18, TBC1D20

□ **comprehensive diagnostic see [GP032XL]**

☐ **Pontocerebellar hypoplasia [GP033] (20,2 kb)**

CASK, RARS2, RELN, TSEN2, TSEN34, TSEN54, VRK1

☐ **Malformations of cortical development (MCD) [GP032XL]**

ACTB, ACTG1, AKT3, AP4M1, ARFGEF2, ARX, ASPA, ASPM, , B3GNT1, CASK, CDK5RAP2, CENPJ, CEP135, CEP152, CEP63, COL18A1, COL4A1, CPT2, DCX, DHCR24, DNM1L, DYNC1H1, EFTUD2, EMX2, EOMES, EXOSC3, EZH2, FGFR3, FH, FKRP, FKTN, FLNA, GCDH, GFAP, GPC3, GPR56, HEPACAM, IER3IP1, ISPD, KIF11, LAMA2, LAMB2, LAMC3, LARGE, MCPH1, MED12, MEF2C, MLC1, MRE11A, MSMO1, NDE1, NFIX, NHEJ1, NR2E1, NSD1, OCLN, OPHN1, PAFAH1B1, PAX6, PEX7, PIGA, PIGN, PIK3CA, PIK3R2, PNKP, POMGNT1, POMT1, POMT2, PQBP1, PTEN, RAB18, RAB3GAP1, RAB3GAP2, RARS2, RELN, SEPSECS, SLC25A19, SNAP29, SRPX2, STIL, TSEN2, TSEN34, TSEN54, TUBA1A, TUBA8, TUBB2B, TUBB3, TUBGCP6, VLDLR, VRK1, WDR62, YWHAE

☐ **Makrocephaly [GP030XL]**

ASPA, BRAF, BRWD3, DHCR24, EZH2, GCDH, GFAP, GPC3, HEPACAM, HRAS, KIF7, MED12, MLC1, NF1, NFIX, NSD1, PIK3CA, PIK3R2, PTEN, SPRED1

☐ **Adams-Oliver syndrome [GP024] (~16 kb)**

ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ

☐ **Aicardi-Goutières syndrome (AGS) [GP025] (~12 kb)**

ADAR, IFIH1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1

☐ **Joubert syndrome [GP004] (~25 kb)**

AHI1, CC2D2A, CEP290, NPHP RPGRIP1L, TMEM67

□ **comprehensive diagnostic see [GP004XL]**

AHI1, ARL13B, B9D1, C5orf42, CC2D2A, CEP290, CEP41, CSPP1, KIF7, MKS1, NPHP1, OFD1, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM237, TMEM67, TTC21B

☐ **Zellweger syndrome [GP035] (~18 kb)**

1. PEX1, 2. PEX6, 3. ABCD3, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5

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Neurological diseases

Movement disorders

☐ **Ataxia and related [GP109XL]**

ABCB7, ABHD12, AFG3L2, ANO10, APTX, ATCAY, ATM, ATP1A3, C10orf2, CA8, CACNA1A, CCDC88C, DNMT1, ELOVL4, FGF14, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, MRE11A, MTPAP, PDYN, PEX10, PEX2, PLEKHG4, POLG, PRKCG, SACS, SETX, SIL1, SLC1A3, SPG7, SPTBN2, SYNE1, SYT14, TDP1, TGM6, TSEN54, TTBK2, TTPA, VLDLR, WFS1

The panel does not include analysis for repeat extensions.

☐ **Charcot-Marie-Tooth disease, demyelinating (CMT1) [GP110] (~25 kb)**

1. PMP22 MLPA, 2. PMP22, DNM2, EGR2, FGD4, FIG4, GDAP1, GJB1, LITAF, MPZ, MTMR2, NEFL, PRX, SH3TC2

☐ **comprehensive diagnostic [GP111XL]**

☐ **Charcot-Marie-Tooth disease, axonal (CMT2) [GP111] (~24 kb)**

DNM2, GAN, GARS, GDAP1, GJB1, HSPB1, HSPB8, IGHMBP2, INF2, MFN2, MPZ, NEFL, TRPV4

☐ **comprehensive diagnostic [GP111XL]**

AARS, ARHGEF10, DNAJB2, DNM2, DYNC1H1, GAN, GARS, GDAP1, GJB1, HSPB1, HSPB8, IGHMBP2, INF2, KIF1B, LMNA, LRSAM1, MED25, MFN2, MPZ, NAGLU, NEFL, PRPS1, RAB7A, SPTLC1, TRPV4, YARS

☐ **Hereditary spastic paraplegia (HSP) [GP114XL]**

1. SPAST, ATL1, 2. BSCL2, CYP7B1, FA2H, GJC2, HSPD1, KIAA0196, KIF5A, NIPA1, PNPLA6, REEP1, RTN2, SLC33A1, SPG11, SPG20, SPG21, SPG7, ZFYVE27

☐ **uncomplicated Form [GP114-aXL]**

1. SPAST, ATL1, 2. AP5Z1, CYP7B1, HSPD1, KIAA0196, KIF5A, NIPA1, REEP1, RTN2, SLC33A1, SPG11, SPG7, ZFYVE27

☐ **complicated Form [GP114-bXL]**

AP4B1, AP4M1, AP4S1, BSCL2, C12ORF65, C19ORF12, CYP7B1, ERLIN2, FA2H, GAD1, GJC2, KIF1A, KIF5A, L1CAM, PLP1, PNPLA6, REEP1, SLC16A2, SPG11, SPG20, SPG21, SPG7, ZFYVE26

☐ **Dystonia [GP113] (~ 22kb)**

ADCY5, ATP1A3, GCH1, GNAL, PNKD, PRRT2, SGCE, SLC2A1, SPR, TAF1, TH, THAP1, TOR1A

Neurodegenerativ diseases

☐ **Neurodegeneration with brain iron accumulation (NBIA) [GP121XL]**

1. PANK2, 2. ATP13A2, C19orf12, COASY, CP, DCAF17, FA2H, FTL, PLA2G6, WDR45

☐ **Parkinson disease [GP118] (~25 kb)**

ATP13A2, DJ1 (PARK7), LRRK2, PARKIN (PARK2), PINK1, SNCA, VPS35

☐ **comprehensive diagnostic [GP118XL]**

ATP13A2, ATP1A3, ATP6AP2, ATXN2, ATXN3, COMT, DCTN1, DNAJC6, EIF4G1, FBXO7, FTL, GBA, GCH1, HTRA2, LRRK2, MAPT, PARK2, PARK7, PINK1, PLA2G6, POLG, PRKRA, SLC30A10, SLC6A3, SNCA, SPR, TAF1, TH, VPS35

☐ **Refsum syndrome [GP010] (~8 kb)**

PEX1, PEX2, PEX26, PEX7, PHYH

☐ **Alzheimer and dementia [GP134] (~21 kb)**

1. PSEN1, APP, PSEN2
2. C9orf72, CHMP2B, FUS, GRN, ITM2B, MAPT, PRNP, SIGMAR1, TARDBP, TREM2, UBQLN2, VCP

☐ **Frontotempora dementia (FTD) [GP135] (~21 kb)**

1. MAPT, GRN
2. CHMP2B, FUS, TARDBP, VCP
3. APP, ITM2B, PRNP, PSEN1, PSEN2, SIGMAR1, TREM2, UBQLN2

☐ Repeatveränderungen für *C9orf72*

☐ **Cerebral microangiopathy (CADASIL, Fabry disease...) [GP158] (~20 kb)**

1. NOTCH3, 2. COL4A1, COL4A2, GLA, HTRA1

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Muscular diseases

☐ **Amyotroph lateral sclerosis (ALS) [GP122XL]**

1. SOD1, C9orf72, 2. ALS2, ANG, CHMP2B, DCTN1, ERBB4, FIG4, FUS, MATR3, NEFH, OPTN, PFN1, PRPH, SETX, SQSTM1, TARDBP, UBQLN2, VAPB, VCP, VEGFA

☐ **Emery-Dreyfuss muscle dystrophy [GP123XL]**

EMD, FHL1, LMNA, SYNE1, SYNE2

☐ **Limb-girdle muscular dystrophy (LGMD) [GP124] (~16 kb)**

1. SGCA, SGCB, SGCG, SGCD, 2. ANO5, CAPN3, DYSF, FKRP

☐ **Muscular dystrophies [GP125XL]**

ANO5, B3GNT1 (=B4GAT1), CAPN3, CHKB, COL6A1, COL6A2, COL6A3, DMD, DYSF, EMD, FHL1, FKRP, FKTN, ISPD, LAMA2, LARGE, LMNA, PABPN1, POMGNT1, POMT1, POMT2, SEPN1, SGCA, SGCB, SGCD, SGCG, SYNE1, SYNE2, TCAP, TTN

☐ **Walker-Warburg syndrome [GP133] (~16kb)**

B3GNT1 (=B4GAT1), FKRP, FKTN, ISPD, LARGE, POMGNT1, POMT1, POMT2, TCAP, TRIM32

☐ **Myasthenic syndrome, congenital [GP126] (~20 kb)**

1. CHRNE, 2. COLQ, DOK7, RAPSN, CHAT, GFPT1, 3. AGRN, CHRNA1, CHRNB1, CHRND

- ☐ **comprehensive diagnostic [GP126XL]**

  AGRN, ALG2, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COLQ, DOK7, DPAGT1, FBN2, GFPT1, LAMB2, LRP4, MUSK, PLEC, PREPL, RAPSN, SCN4A, TPM2

☐ **Myopathy, myofibrillar (MFM) [GP128] (~16 kb)**

DES, CRYAB, MYOT, LDB3, FLNC, BAG3, FHL1, DNAJB6

☐ **Myopathy, nemaline (NM) [GP129XL]**

1. NEB (25 kb), ACTA1, 2. TPM3, TPM2, TNNT1, CFL2

☐ **Myopathy, comprehensive panel [GP130XL]**

ACTA1, ACVR1, ANO5, BAG3, BIN1, C10orf2, CAV3, CFL2, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, CRYAB, DES, DNM2, DYSF, FHL1, FKBP14, FLNC, GNE, ISCU, KBTBD13, KLHL9, LAMP2, LDB3, MAMLD1, MATR3, MEGF10, MSTN, MTM1, MTMR14, MYF6, MYH14, MYH2, MYH7, MYOT, NEB, OPA1, ORAI1, PABPN1, PLEC, POLG, POLG2, PUS1, RRM2B, RYR1, SEPN1, SIL1, STIM1, SUCLA2, TK2, TNNT1, TPM2, TPM3, TRIM32, TTN, VCP, VMA21, YARS2

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Bone- and skeletal diseases

☐ **3M-Syndrom [GP048] (~12 kb)**
CUL7, OBSL1, CCDC8

☐ **Adams-Oliver syndrome [GP024] (~16 kb)**
ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ

☐ **Arthrogryposis multiplex congenita (AMC) [GP049XL]**
ECEL1, FBN2, MYBPC1, MYH3, MYH8, PIEZO2, TNNI2, TNNT3, TPM2

☐ **Chondrodysplasia punctata [GP050] (~7 kb)**
AGPS, ARSE, EBP, GNPAT, PEX7

☐ **Klippel-Feil syndrome [GP169] (~13 kb)**
GDF3, GDF6, MEOX1, MYO18B, PAX1, RIPPLY2

☐ **Multiple epiphysale Dysplasie [GP051] (~18 kb)**
1. COMP, 2. COL2A1, COL9A1, COL9A2, COL9A3, MATN3, SLC26A2

☐ **Metaphyseal Dysplasia [GP055]** (~10 kb)
COL10A1, MMP9, MMP13, PTH1R, RMRP, RUNX2, SBDS

☐ **Osteogenesis imperfecta [GP057]** (~24 kb)
1. COL1A1, 2. COL1A2, 3. BMP1, CRTAP, FKBP10, IFITM5, LEPRE1 (P3H1), PLOD2, PPIB, SERPINF1, SERPINH1, SP7

☐ **Radial malformations [GP058] (~9 kb)**
SALL4, TBX5, SALL1

Noonan syndrome und RASopathies [GP092]

☐ **Noonan syndrome [GP092a]** (nach EBM 11355 und 11356)
1. PTPN11
2. BRAF, KRAS, NRAS, PIK3CA, PTPN11, RAF1, RASA1, RIT1, SOS1

☐ **RASopathies [GP092b]** (without genes for Noonan syndrome) (~17kb)
CBL, HRAS, MAP2K1, MAP2K2, NF1, SHOC2, SPRED1

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Skin and connective tissue

☐ **Albinism, oculocutaneous [GP168] (~10 kb)**
MC1R, OCA2, SLC24A5, SLC45A2 (MATP), TYR, TYRP1

☐ **Aortic aneurysm and dissection (TAAD) [GP037]**
1. COL3A1, 2. ACTA2, CBS, COL3A1, COL5A1, COL5A2, FBN1, FBN2, FLNA, MED12, MYH11, MYLK, NOTCH1, SKI, SLC2A10, SMAD3, SMAD6, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2

☐ **Ehlers-Danlos syndrome (EDS) [GP176XL]**
ADAMTS2, COL3A1, COL5A1, COL5A2, COL1A1, COL1A2, PLOD1, TNXB

☐ **Marfan syndrome [GP044]** *(EBM11445, 11446, 11448)*
1. FBN1, 2. TGFBR1, TGFBR2
ggfs. ACTA2, FBN1, MYH11, MYLK, SMAD3, TGFB2

☐ **Cutis Laxa [GP161]** (~25 kb)
ALDH18A1, ATP6V0A2, EFEMP2, ELN, FBLN5, GORAB, LTBP4, PYCR1

☐ **Epidermolysis bullosa (EB) [GP041XL, 28,8kb]**
COL17A1, COL7A1, KRT14, KRT5, LAMA3, LAMB3, LAMC2

☐ **Ichthyosis, autosomal recessive (ARCI) [GP043] (~21 kb)**
1. TGM1 2. ABCA12, ABHD5, ALOX12B, ALOXE3, CERS3, CYP4F22, NIPAL4, PNPLA1

☐ **Genodermatosis, comprehensive panel [GP163XL]**
AAGAB, ABCA12, ABHD5, ABHD5, ALDH3A2, ALOX12B, ALOXE3, AP1S1, AQP5, CERS3, CLDN1, CYLD, CYP4F22, DSG, DSG1, DSP, ENPP1, SLC27A4 (FATP4), GJA1, GJB3, GJB4, IL36RN, KRT1, KRT10, KRT14, KRT16, KRT16, KRT17, KRT5, KRT9, LOR, MBTPS2, NIPAL4, PNPLA1, POMP, PORCN, RASA1, RECQL4, SLURP1, SPINK5, STS, TGM1, TGM5, TRPV3

☐ **Dyskeratosis congenita (DC, DKC) [GP038] (~kb)**
CTC1, DKC1, NHP2, NOP10, RTEL1, TERC, TERT, TINF2, WRAP53

☐ **Ectodermal dysplasia (ED) [GP040XL]**
ANTXR1, APCDD1, AXIN2, BANF1, BCS1L, CDH3, CDSN, CTSC, DLX3, DSG4, DSP, EDA, EDAR, EDARADD, FGF10, FGFR2, FGFR3, GJA1, GJB6, GRHL2, GTF2H5, HR, IFT122, IFT43, KRT14, KRT74, LIPH, LPAR6, MPLKIP, MSX1, NFKBIA, OFD1, PIGL, PKP1, PORCN, PVRL1, PVRL4, SHOC2, SOX18, ST14, TP63, TRPS1, TWIST2, UBR1, WDR19, WDR35, WNT10A

☐ **Stickler syndrome [GP013] (~22 kb)**
COL11A1, COL2A1, COL9A2, COL9A3, COL11A2, COL9A1

☐ **Fanconi anemia [GP042] (~24 kb)**
BRCA2, FANCA, FANCC, FANCG, FANCD2, FANCE

- ☐ **comprehensive diagnostic [GP042XL]**
  BRCA2, BRIP1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, PALB2, RAD51C, SLX4

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Throat, nose and ear

☐ **Alport syndrome [GP062] (~25 kb)**

1. COL4A5, 2.COL4A3, COL4A4, COL4A6, MYH9

☐ **Hypogonadism / Kallmann syndrome [GP098]**

AXL, CHD7, FEZF1, FGFR1, FGF8, FGF17, HS6ST1, IL17RD, KAL1, HESX1, NSMF, PROK2, PROKR2, SEMA3A, SOX10, WDR11

☐ **comprehensive diagnostic [GP098XL]**

CHD7, DMP1, DUSP6, FEZF1, FGF17, FGF23, FGF8, FGFR1, FLRT3, GNRH1, GNRHR, HS6ST1, IL17RD, KAL1, KISS1R, NELF, NSMF, PHEX, PROK2, PROKR2, SEMA3A, SPRY4, TAC3, TACR3, WDR11

☐ **Refsum syndrome [GP010] (~7 kb)**

PEX1, PEX2, PEX26, PEX7, PHYH

☐ **Nonsyndromic hearing loss and deafness [GP064] (~25 kb)**

GJB2, GJB6, MYO7A, POU3F4, SLC26A4, USH2A

☐ **comprehensive diagnostic [GP064XL]**

ACTG1, CCDC50, CDH23, CEACAM16, CIB2, CLDN14, COCH, COL11A2, COL4A6, CRYM, DFNA5, DFNB31, DFNB59, DIABLO, DIAPH1, DIAPH3, DSPP, ESPN, ESRRB, EYA4, GIPC3, GJB2, GJB3, GJB6, GPSM2, GRHL2, GRXCR1, HGF, ILDR1, KARS, KCNQ4, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MIR96, MSRB3, MYH14, MYH9, MYO15A, MYO1A, MYO3A, MYO6, MYO7A, OTOA, OTOF, PCDH15, POU3F4, POU4F3, PRPS1, PTPRQ, RDX, SERPINB6, SLC17A8, SLC26A4, SLC26A5, SMPX, STRC, TECTA, TJP2, TMC1, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, USH1C, USH2A, WFS1

☐ **Syndromic hearing loss and deafness [GP065XL]**

ABHD12, ALMS1, ANKH, ATP6V1B1, BSND, CACNA1D, CD151, CDH23, CDKN1C, CHD7, CHSY1, CIB2, CLDN14, CLRN1, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, DFNB31, DFNB59, DLX5, EDN3, EDNRB, ESPN, ESRRB, EYA1, FGF3, FOXI1, GATA3, GIPC3, GJB2, GJB3, GJB6, GPR98, GPSM2, GRXCR1, HGF, ILDR1, KCNE1, KCNJ10, KCNQ1, LHFPL5, LOXHD1, LRTOMT, MANBA, MARVELD2, MITF, MSRB3, MYO15A, MYO3A, MYO6, MYO7A, NDP, NLRP3, OTOA, OTOF, PAX3, PCDH15, PDZD7, POLR1C, POLR1D, POU3F4, RDX, SEMA3E, SIX1, SIX5, SLC12A1, SLC19A2, SLC26A4, SLC26A5, SMPX, SNAI2, SOX10, STRC, TCOF1, TECTA, TFAP2A, TIMM8A, TMC1, TMIE, TMPRSS3, TPRN, TRIOBP, TYR, USH1C, USH1G, USH2A, WFS1

☐ **Stickler syndrome [GP013] (~22 kb)**

COL11A1, COL2A1, COL9A2, COL9A3, COL11A2, COL9A1

☐ **USHER syndrome [GP091] (~24 kb)**

1. USH2A, 2. MYO7A, USH1C

☐ **comprehensive diagnostic [GP091XL]**

CDH23, CIB2, CLRN1, DFNB31, HARS, MYO7A, PCDH15, PDZD7, USH1C USH1C, USH1G, USH2A

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Cardiological diseases

☐ **Thoracic aortic aneurysm and dissection (TAAD) [GP037XL]**

1. COL3A1, 2. ACTA2, CBS, COL3A1, COL5A1, COL5A2, FBN1, FBN2, FLNA, MED12, MYH11, MYLK, NOTCH1, SKI, SLC2A10, SMAD3, SMAD6, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2

☐ **Congenital cardiac defects [GP173] (~ 25 kb)**

ACTC1, CITED2, FOXH1, FOXP1, GATA5, GATA4, GATA6, GJA1, MYH6, NKX2-5, TBX1, TBX20

☐ **Brugada syndrome [GP084] (~28 kb)**

1. SCN5A, 2. SCN10A, CACNA1C, TRPM4, CACNB2, SCN1B, CACNA2D1

☐ **Long-QT syndrome [GP090] (~24 kb)**

1. KCNQ1, KCNH2, 2. SCN5A, ANK2, KCNE2

□ **comprehensive diagnostic [GP090XL]**

AKAP9, ANK2, CACNA1C, CALM1, CAV3, DSP, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1

☐ **Short-QT syndrome and atrialfibrillation [GP094] (~23 kb)**

ABCC9, GJA5, KCNA5, KCNE1, KCNH2, KCNJ2, KCNQ1, NPPA, SCN2B, SCN3B, SCN4B, SCN5A

☐ **Cardiac arrhythmia, general [GP159XL]**

ABCC9, AKAP9, ANK2, CACNA1C, CACNA2D1, CACNB2, CALM1, CAV3, DSP, GJA5, KCNA5, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, NPPA, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SNTA1, TRPM4

☐ **Cardiomyopathy, arrhythmogenic right ventricular (dysplasia) (ARVD/C) [GP082] (~20 kb)**

1. DSG2, DSP, DSC2, PKP2, JUP

□ **comprehensive diagnostic [GP082XL]**

DES, DSC2, DSG2, DSP, JUP, LMNA, PKP2, PLN, RYR2, TGFB3, TMEM43, TTN

☐ **Cardiomyopathy, dilated [GP085XL]**

ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, CRYAB, CSRP3, DES, DMD, DSG2 , DSP , EMD, EYA4, FKTN, GATAD1, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYPN, NEBL, NEXN, PDLIM3, PLN, PSEN1, RAF1, RBM20, SCN5A, SGCD, TAZ, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTN, VCL

☐ **Cardiomyopathy, hypertrophic [GP087] (~25 kb)**

1. MYH7, MYBPC3, 2. ACTC1, ACTN2, CSRP3, MYH6, MYL2, MYL3, MYOZ2, NEXN, PLN, TNNT2, TCAP, TNNC1, TNNI3

☐ **Cardiomyopathy,left ventricular noncompaction (LVNC) [GP089] (~25 kb)**

ACTC1, DTNA, LDB3, LMNA, MIB1, MYBPC3, MYH7, PRDM16, TAZ, TNNT2, TPM1

Noonan syndrome and RASopathies [GP092]

☐ **Noonan syndrome [GP092a]** (nach EBM 11355 und 11356)

1. PTPN11
2. BRAF, KRAS, NRAS, PIK3CA, PTPN11, RAF1, RASA1, RIT1, SOS1

☐ **RASopathies [GP092b]** (without genes for Noonan syndrome) (~17kb)

CBL, HRAS, MAP2K1, MAP2K2, NF1, SHOC2, SPRED1

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Liver, kidney, endocrinology

☐ **Alport syndrome [GP062] (~25 kb)**

1. COL4A5 2.COL4A3, COL4A4, COL4A6, MYH9

☐ **Bardet-Biedl syndrome [GP003] (~20 kb)**

ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, MKKS, MKS1, TTC8

☐ **comprehensive diagnostic [GP003XL]**

ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CCDC28B, CEP290, LZTFL1, MKKS, MKS1, NPHP1, SDCCAG8, TMEM67, TRIM32, TTC21B, TTC8, WDPCP

☐ **Bartter syndrome [GP096] (~16 kb)**

BSND, CASR, CLCNKA, CLCNKB, KCNJ1, SLC12A1, SLC12A3

☐ **Focal segmental glomerulosclerosis (FSGS) [GP097] (~18 kb)**

ACTN4, ARHGAP24, CD2AP, INF2, MYO1E, PAX2, TRPC6

☐ **Hypogonadism / kallmann syndrome [GP098] (~x kb)**

AXL, CHD7, FEZF1, FGFR1, FGF8, FGF17, HS6ST1, IL17RD, KAL1, HESX1, NSMF, PROK2, PROKR2, SEMA3A, SOX10, WDR11

☐ **comprehensive diagnostic [GP098XL]**

CHD7, DMP1, DUSP6, FEZF1, FGF17, FGF23, FGF8, FGFR1, FLRT3, GNRH1, GNRHR, HS6ST1, IL17RD, KAL1, KISS1R, NELF, NSMF, PHEX, PROK2, PROKR2, SEMA3A, SPRY4, TAC3, TACR3, WDR11

☐ **Familial hyperinsulinism (FHI) [GP160] (~18 kb)**

1. ABCC8, 2. GCK, GLUD1, HADH, HNF4A, INSR, KCNJ11, SLC16A1 UCP2

☐ **Joubert syndrome [GP004] (~25 kb)**

AHI1, CC2D2A, CEP290, NPHP RPGRIP1L, TMEM67

☐ **comprehensive diagnostic [GP004XL]**

AHI1, ARL13B, B9D1, C5orf42, CC2D2A, CEP290, CEP41, CSPP1, KIF7, MKS1, NPHP1, OFD1, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM237, TMEM67, TTC21B

☐ **Meckel syndrome [GP100XL]**

B9D1, B9D2, CC2D2A, CEP290, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM216, TMEM67

☐ **Nephronophthisis [GP102] (~25 kb)**

CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, TMEM67

☐ **comprehensive diagnostic [GP102XL]**

CEP290, DCDC2, GLIS2, INVS, IQCB1, NEK8, NPHP1, NPHP3, NPHP4, SLC41A1, TMEM67, TTC21B, WDR19, XPNPEP3

☐ **Polycystic kidney disease (PKD) [GP103XL]**

1. PKD1, PKD2, PKHD1 (~28 kb)
2. HNF1B, MUC1, PAX2, UMOD, BICC1 (~9 kb)

☐ **Premature ovarian failure (POI, POF) [GP104] (~14 kb)**

AMH, BMP15, FIGLA, FOXL2 FSHR, INHA, NOBOX, NR5A1, PGRMC1 TGFBR3
Additional genetic tests
☐ *FMR1 (FraX)*1*
☐ *Chromosomal testing (Li-Heparin blood) *1*

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Metabolism and hematopoiesis

Metabolism

☐ **Maple syrup urine disease (MSUD) [GP137] (~5kb)**

1. BCKDHA, 2. BCKDHB, 3. DBT, PPM1K

☐ **Familial hyperinsulinism (FHI) [GP160] (~18)**

1. ABCC8, 2. GCK, GLUD1, HADH, HNF4A, INSR, KCNJ11, SLC16A1 UCP2

☐ **Glycogen storage disease (CDG) [GP086XL] (30kb)**

AGL, G6PC, GAA, GBE1, GYS2, LAMP2, PFKM, PHKA2, PHKB, PHKG2, PYGL, PYGM, SLC37A4

☐ **Glycosylation disorders (CDG) [GP079] (~22 kb)**

1. PMM2, 2. MPI, ALG6, 3. ALG1, ALG12, ALG2, ALG8, ALG9, DOLK, DPAGT1, DPM1, MAGT1, MPDU1, RFT1 SRD5A3, TMEM165, TUSC3

☐ **Hereditary hemochromatosis [GP175] (7 kb)**

FTL, HFE, HFE2, HAMP, TFR2, SLC40A1

☐ **Maturity-onset diabetes of the young (MODY) [GP140] (20,9kb)**

ABCC8, BLK, CEL, GCK, HNF1A, HNF1B (=TCF2), HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1

☐ **Mucopolysaccharidosis (MPS) [GP141]**

ARSB, GALNS, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, LDB3, MYOT, NAGLU, SGSH

☐ **Neuronal ceroid lipofuscinoses (NCL) [GP142XL]**

ASAH1, ATP13A2, CLN3, CLN5, CLN6, CLN8, CTSD, DNAJC5, GRN, KCTD7, MFSD8, NHLRC1, PPT1, TPP1

☐ **Zellweger syndrome [GP035] (~18 kb)**

1. PEX1, 2. PEX6, 3. ABCD3, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5

Hematopoiesis

☐ **Bernard-Soulier syndrome [GP076]**

GP1BA, GP1BB, GP9

☐ **Hermansky-Pudlak syndrome [GP167] (~22 kb)**

AP3B1, AP3D1, BLOC1S3, BLOC1S6, DTNBP1, HPS1 HPS3, HPS4, HPS5, HPS6, PLDN

☐ **Fanconi Anemia [GP042] (~24 kb)**

BRCA2, FANCA, FANCC, FANCG, FANCD2, FANCE

☐ **comprehensive diagnostic [GP042XL]** (28,9kb)

BRCA2, BRIP1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, PALB2, RAD51C, SLX4

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Mitochondria diseases

☐ **MELAS syndrome [GP164]**

MT-TL1, MT-ND1, MT-ND5, MT-ND4, MT-ND6, MT-CYB, MT-CO3, MT-CO1, MT-CO2, MT-ND3, MT-TF, MT-TQ, MT-TH, MT-TV, MT-TW, MT-TK, MT-TL2, MT-TE, MT-TC, MT-TS1, MT-TS2

☐ **Liver hereditary optic neuropathy (LHON) [GP165]**

MT-ATP6, MT-CO1, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT - ND4, MT-ND4L MT-ND5, MT-ND6, MT-RNR1, MT-TE, MT-TL1, MT-TM, MT - TQ, MT- TT

☐ **Mitochondrial myopathy / encephalopathy [GP166]**

MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND2, MT-ND5, MT-RNR2 MT-TA, MT- TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT - TL1, MT-TL2, MT- TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TW

☐ **Mitochondrial genome [GP105]**

MT-ND4, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT- TW, MT-TY

☐ **Nuclear encoded, mitochondrial genes [GP106XL]**

AARS2, ABCB7, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, ADCK3, AFG3L2, AGK, AIFM1, AK2, ALAS2, ALDH4A1, ALDH6A1, AMACR, AMT, APTX, ATL1, ATP5E, ATPAF2, AUH, BCAT2, BCKDHA, BCKDHB, BCS1L, BOLA3, BTD, C10orf2, C12orf65, CISD2, COA5, COQ2, COQ6, COQ9, COX10, COX14, COX15, COX4I2, COX6B1, CPS1, CPT1A, CPT2, CYB5R3, CYCS, CYP27A1, D2HGDH, DARS2, DBT, DECR1, DGUOK, DHODH, DIABLO, DLAT, DLD, DMGDH, DNAJC19, DNM1L, EARS2, ELAC2, ERCC6, ETFA, ETFB, ETFDH, ETHE1, FARS2, FASTKD2, FBP1, FH, FOXRED1, FXN, GAMT, GARS, GATM, GCDH, GCK, GCSH, GDAP1, GFER, GFM1, GK, GLDC, GLRX5, GLUD1, HADH, HADHA, HADHB, HARS2, HCCS, HIBCH, HK1, HLCS, HMGCL, HMGCS2, HOGA1, HSD17B10, HSPD1, HTRA2, IDH1, IDH2, IDH3B, ISCU, IVD, KARS, KIF1B, KIF5A, L2HGDH, LARS2, LIAS, LRPPRC, MAOA, MARS2, MCCC1, MCCC2, MCEE, MFN2, MLYCD, MMAA, MMAB, MMADHC, MPV17, MRPL3, MRPS16, MRPS22, MTFMT, MTO1, MTPAP, MUT, NAGS, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NUBPL, OAT, OPA1, OPA3, OTC, OXCT1, PANK2, PARK2, PARK7, PC, PCCA, PCCB, PCK2, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PINK1, PNKD, PNPLA2, POLG, POLG2, PPOX, PUS1, RARS2, REEP1, RMRP, RRM2B, SACS, SAMHD1, SARS2, SCO1, SCO2, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD, SLC19A2, SLC19A3, SLC22A5, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC25A38, SLC25A4, SLC33A1, SLC6A8, SPAST, SPG20, SPG7, STAR, SUCLA2, SUCLG1, SURF1, TACO1, TAZ, TIMM8A, TK2, TMEM126A, TMEM70, TPK1, TRMU, TSFM, TTC19, TUFM, TYMP, UNG, UQCRB, UQCRQ, WFS1, WWOX, XPNPEP3, YARS2, YWHAE

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Cancer syndromes (germline)

☐ **Breast cancer, hereditary [GP148]**

1: BRCA1, BRCA2,
2. CHEK2, PALB2, RAD51C
3. **comprehensive diagnostic [GP148XL]**
ATM, BARD1, BLM, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, FAM175A, MEN1, MRE11A, NBN, PALB2, PTEN, RAD50, RAD51C, RAD51D, STK11, TP53

☐ **Ovarian cancer [GP151XL]**

BARD1, BRCA1, BRCA2, BRIP1, MLH1, MRE11A, MSH2, NBN, PALB2, PMS2, RAD50, RAD51C, RAD51D, TP53

☐ **Fanconi anemia [GP042] (~24 kb)**

BRCA2, FANCA, FANCC, FANCG, FANCD2, FANCE

☐ **comprehensive diagnostic [GP042XL]**

BRCA2, BRIP1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, PALB2, RAD51C, SLX4

☐ **Gastrointestinal stroma tumors (GIST) [GP143] (~19 kb)**

KIT, MAX, MEN1, NF1, PRKAR1A, SDHB, SDHC, SDHD, SMARCB1, TMEM127

☐ **Gorlin-Goltz syndrome [GP144] (~10 kb)**

PTCH1, PTCH2, SUFU

☐ **Hereditary nonpolyposis colorectal cancer (HNPCC) / Lynch syndrome [GP145]** (nach EBM 11431 od. 11432)

MLH1, MSH2, MSH6, PMS2, EPCAM (nur Deletionen)

☐ **Colon cancer [GP146XL]**

APC, BMPR1A, BUB1B, MET, CHEK2, MLH1, MSH3, MSH6, MUTYH, NBN, PMS1, PMS2, PTEN, SMAD4, STK11, TP53

☐ **Gastric carcinoma [GP147] (~23 kb)**

BMPR1A, CDH1, CHEK2, MLH1, MSH2, MSH6, PMS2, STK11, TP53

☐ **Melanoma, hereditary [GP149] (~9,5 kb)**

CDK4, CDKN2A, MC1R, MITF, POT1, PTEN, RB1

☐ **Renal carcinoma [GP150XL]**

CHEK2, EPCAM (nur Deletionen), FH, FLCN, GPC3, HRPT2, MET, MLH1, MSH2, MSH6, PMS2, PTEN, SDHB, SDHC, SDHD, SMARCB1, TP53, TSC1, TSC2, VHL, WT1

☐ **Pancreatic cancer (including chronic pancreatitis) [GP152XL]**

APC, ATM, BRCA1, BRCA2, CDK4, CDKN2A, MLH1, MSH2, MSH6, PALB2, PMS2, PRSS1, SPINK1, STK11, TP53

☐ **Paraganglioma - pheochromocytoma syndromes [GP153] (~19 kb)**

MAX, MEN1, NF1, PRKAR1A, RET, SDHB, SDHC, SDHD, TMEM127, VHL

☐ **Polyposis coli [GP157XL]**

APC, BMPR1A, CHEK2, FLCN, MSH3, MUTYH, NTHL1, POLD, POLE, PTEN, SMAD4, STK11

☐ **Thyroid carcinoma [GP154] (~6 kb)**

PTEN, RET, SDHB, SDHC, SDHD

☐ **Cancer snydromes, extended panel [GP155XL]**

APC, AR, ATM, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BTNL2, BUB1B, CDH1, CDK4, CDKN2A, CHEK2, ELAC2, FH, FLCN, GPC3, HOXB13, KIT, MAX, MC1R, MEN1, MET, MITF, MLH1, MRE11A, MSH2, MSH6, MSR1, MUTYH, NBN, NF1, PALB2, PMS1, PMS2, PRKAR1A, PTEN, RAD50, RAD51C, RAD51D, RB1, RET, RNASEL, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCB1, STK11, TMEM127, TP53, TSC1, TSC2, VHL, WT1, ZFHX3

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