



Leistungsangebot: Syndromologie und Epilepsien

Hinweise:

- Durchführung der Analysen mittels NGS. Triplet-Repeats, grössere Deletionen/Duplikationen und grobstrukturelle, chromosomale Veränderungen werden nicht erfasst
- Bei Verordnung dieser Analysen sollte eine Kostengutsprache vom anfordernden Arzt eingeholt werden. Wir unterstützen Sie gerne dabei.
- Verordnung von Genpanels mit über 10 Genen gemäss AL des BAG nur durch Ärzte mit dem Titel FMH Medizinische Genetik.
- Die hier gelisteten Genpanels stellen nur einen Auszug aus den möglichen Analysen dar. Sie können auf die Fragestellung spezifisch/individuell angepasst werden
- Die Zusammenstellung der Genpanels kann abhängig von der Datenlage und dem kommerziellen Anbieter abweichen.

□ Noonan Syndrom (17 Gene) [EPSY01]

BRAF, KRAS, NRAS, PTPN11, RAF1, RIT1, SOS1, LZTR1, MAP2K1, MRAS, RRAS2, SHOC2, SOS2, CBL, MAP2K2, PPP1CB, HRAS

□ Rasopathien erweitert (21 Gene) [EPSY02]

BRAF, CBL, FGD1, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NF1, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1, RASA2

□ Epilepsie (häufige und therapierelevante Gene) (24 Gene) [EPSY03]

ALDH4A1, ALDH7A1, ALPL, ARX, CDKL5, FOLR1, GABRA1, GAMT, GRIN2A, KCNQ2, KCNT1, PCDH19, PLCB1, PLPBP, PNPO, PRRT2, SCN1A, SCN2A, SCN8A, SLC2A1, SLC35A2, SLC6A8, STXBP1, TPP1

□ Epilepsie erweitert (271 Gene oder Genauswahl nach Absprache) [EPSY04]

AARS, ABAT, ACTL6B, ACY1, ADAM22, ADGRV1, ADK, ADRA2B, ADSL, ALDH7A1, ALG13, AFG3L2, AP3B2, ALDH5A1, ALDH7A1, ARHGEF15, ARHGEF9, AMT, ANKRD11, AP3B2, ARHGEF9, ARV1, ARX, ASAH1, ATAD1, ATIC, ATP13A2, ATP1A2, ATP6V1A, AUH, BCKDK, BRAT1, BTB, CACNA1A, CACNA1E, CACNA1H, CACNB4, CAD, CAMK2A, CAMK2B, CASR, CASK, CDKL5, CERS1, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN2, CLCN4, CLN3, CLN5, CLN6, CLN8, CNM2, CNPY3, CNTN2, COQ4, COQ8A, CPA6, CPLX1, CPT1A, CPT2, CSTB, CTSD, CTSF, CUX2, CYFIP2, D2HGDH, DCX, DDX3X, DENND5A, DEPDC5, DHDDS, DHFR, DNAJC5, DNM1, DOCK7, DPYD, DYRK1A, EEF1A2, EFHC1, EPM2A, ETFA, ETFB, ETFDH, ETHE1, FGF12, FOLR1, FOXG1, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAL, GAMT, GATM, GCDH, GCH1, GCSH, GLDC, GLS, GLRA1, GLRB, GLUL, GNAO1, GNB1, GOSR2, GPAA1, GPHN, GRIA4, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRN, GUF1, HACE1, HCN1, HDAC4, HLCS, HNRNPU, HPD, ICK, IDH2, IQSEC2, ITPA, KCNA1, KCNA2, KCNB1, KCNC1, KCNH5, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD7, L2HGDH, LGI1, LIAS, LMNB2, LNP, MBD5, MBOAT7, MDH2, MECP2, MEF2C, MFSD8, MLYCD, MMACHC, MOCS1, MOCS2, MOCS3, MTFMT, MTHFR, MTOR, NECAP1, NEU1, NEXMIF, NEUROD2, NHLRC1, NPRL2, NPRL3, NTRK2, PACS2, PARS2, PC, PCBD1, PCDH19, PDHA1, PDSS2, PGAP1, PGAP2, PGAP3, PHACTR1, PHGDH, PIGA, PIGB, PIGC, PIGG, PIGP, PIGH, PIGL, PIGM, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGV, PIGW, PIGY, PLCB1, PLPBP, PNKP, PNPO, POLG, PPP3CA, PPT1, PRDM8, PRICKLE1, PRICKLE2, PRRT2, PSAT1, PSPH, PTS, PURA, QDPR, RANBP2, RANGAP1, RELN, RHOTB2, RNF13, ROGDI, RORB, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SERPINI1, SIK1, SLC1A3, SLC12A5, SLC13A5, SLC19A3, SLC1A2, SLC25A1, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC46A1, SLC6A1, SLC6A5, SLC6A8, SLC6A9, SMC1A, SPTAN1, SRPX2, ST3GAL3, STX1B, STXBP1, SUOX, SYNGAP1, SYNJ1, SZT2, TBC1D24, TBL1XR1, TCF4, TPP1, TRAK1, TSC1, TSC2, UBA5, UBE3A, WDR45, WWOX, YWHAG, ZEB2



□ **Entwicklungsverzögerung (861 Gene) [EPSY05]**

AARS, AARS2, ABCC9, ABCD1, ACOX1, ACSL4, ACTB, ACTG1, ADAR, ADAT3, ADGRG1, ADNP, ADPRHL2, AFF2, AGA, AGTR2, AHDC1, AHI1, AIFM1, AIMP1, AIMP2, AKT1, AKT2, AKT3, ALDH3A2, ALDH5A1, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ALKBH8, AMACR, AMER1, AMPD2, ANK3, ANKLE2, ANKRD11, AP1S2, AP2M1, APC2, APOPT1, ARF1, ARFGF2, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARID2, ARL13B, ARL3, ARMC9, ARSA, ARSB, ARX, ASH1L, ASNS, ASPA, ASPM, ASXL2, ATP6AP2, ATP7A, ATR, ATRX, AUTS2, B3GALNT2, B4GALT1, B4GAT1, B9D1, B9D2, BCAP31, BCOR, BDNF, BOLA3, BRAF, BRD4, BRWD3, C12orf4, C12orf57, C2CD3, CA8, CACNA1C, CACNG2, CAD, CAMK2A, CAMK2B, CAMK2G, CARS2, CASK, CBL, CC2D1A, CC2D2A, CCDC115, CCDC22, CCDC88C, CCND2, CDH15, CDK5, CDK13, CDK5RAP2, CDK6, CDKL5, CDKN1C, CDON, CENPE, CENPF, CENPJ, CEP104, CEP120, CEP135, CEP152, CEP290, CEP41, CEP63, CHAMP1, CHD3, CHD4, CHD7, CHD8, CHMP1A, CIC, CIT, CLCN4, CLIC2, CLCN2, CLP1, CLTC, CNKSR2, CNOT3, CNTNAP2, COASY, COG1, COG2, COG4, COG5, COG6, COG7, COG8, COL4A1, COL4A2, COL4A3BP, COPB2, COX10, COX15, COX6B1, CPLANE1, CRADD, CRBN, CREBBP, CSF1R, CSNK2A1, CSPP1, CTC1, CTCF, CTNNA2, CTNNB1, CTNS, CTSB, CTU2, CUL4B, CXorf56, CYP27A1, DAG1, DARS, DARS2, DBH, DCHS1, DCX, DDC, DDOST, DDX3X, DEAF1, DHCR24, DHCR7, DHDDS, DIS3L2, DKC1, DLG3, DLG4, DNAJC12, DNM1, DNMT3A, DNMT3B, DOCK7, DOCK8, DOLK, DOLK, DONSON, DPAGT1, DPFB, DPM1, DPM2, DPM3, DPP6, DVL1, DVL3, DYNC1H1, DYRK1A, EARS2, EBP, EDC3, EED, EEF1A2, EFHC2, EHMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF3F, ELP2, EMC1, EMX2, EP300, EPB41L1, EPRS, ERCC6, ERCC8, ERMARD, EXOSC3, EXOSC8, EXOSC9, EYA1, EZH2, FA2H, FAM126A, FANCB, FARS2, FAT4, FBXO31, FGD1, FGFR1, FH, FIBP, FKBP, FKTN, FLNA, FMN2, FMR1, FOLR1, FOXP1, FOXP2, FOXRED1, FRMPD4, FTSJ1, FUCA1, FUT8, GABRA1, GABRA2, GABRG1, GALC, GALNS, GALT, GAN, GATAD2B, GBA, GBE1, GCDH, GCH1, GDI1, GEMIN4, GFAP, GFM1, GIMAP1, GJC2, GK, GLI3, GLA, GLB1, GLI2, GLRX5, GM2A, GMPGA, GMPPB, GNAI1, GNAO1, GNB1, GNPTAB, GNPTG, GNS, GPAA1, GPC3, GPKOW, GPSM2, GPT2, GRIA3, GRIK2, GRIN1, GRIN2A, GRIN2B, GUSB, HCCS, HCFC1, HCN1, HDAC4, HDAC6, HDAC8, HEPACAM, HERC1, HERC2, HEXA, HEXB, HGSNAT, HIKESHI, HIST1H1E, HIVEP2, HMGB3, HNMT, HNRNP2, HPRT1, HRAS, HSD17B10, HSD17B4, HSPD1, HTRA1, HUWE1, HYAL1, IBA57, IDS, IDUA, IER3IP1, IFIH1, IGF2, IGBP1, IKBKG, IL1RAPL1, IMPA1, INPP5E, IQSEC2, IRF6, ISCA1, ISCA2, ISPD, ITPA, KANSL1, KARS, KAT6A, KAT6B, KATNB1, KCNA2, KCNB1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KDM5B, KDM5C, KDM6A, KIAA0442, KIAA0556, KIAA0586, KIF4A, KIF11, KIF14, KIF1A, KIF1BP, KIF2A, KIF4A, KIF5A, KIF5C, KIF7, KIRREL3, KLF8, KLHL15, KMT2A, KMT2D, KMT5B, KNL1, KPTN, KRAS, L1CAM, L2HGDH, LAMA2, LAMB1, LAMC3, LAMP2, LARGE1, LAS1L, LINGO1, LINS1, LIPA, LIPT2, LMAN2L, LMNB1, LRPPRC, LYRM7, LZTR1, MACF1, MAGT1, MAN1B1, MAN2B1, MANBA, MAOA, MAOB, MAP2K1, MAP2K2, MAP2K2, MARS2, MBD5, MBOAT7, MBTPS2, MCOLN1, MCPH1, MECP2, MED12, MED13, MED13L, MED17, MED23, MEF2C, MFSD2A, METTL23, METTL5, MFSD2A, MGAT2, MID1, MID2, MKS1, MLC1, MOGS, MPDU1, MPDZ, MPI, MPV17, MRAS, MSL3, MTFMT, MTHFS, MTM1, MTOR, MYT1L, NAA10, NAA15, NACC1, NAGA, NAGLU, NALCN, NARS2, NAXE, NCAPD2, NCAPD3, NCAPH, NDE1, NDP, NDST1, NDUFA1, NDUFS1, NDUFV1, NEDD4L, NEU1, NEXMIF, NFIB, NFIX, NFU1, NGLY1, NHS, NIN, NIPBL, NKX6-2, NKX6-2, NLGN3, NLGN4X, NONO, NOTCH3, NPC1, NPC2, NPHP1, NRAS, NRXN1, NSD1, NSDHL, NSMCE2, NSUN2, NTNG1, NUBPL, NUP37, NUS1, NXF5, NXN, OCLN, OCRL, OFD1, OGT, OPHN1, OTC, PACS1, PAFAH1B1, PAK1, PAK3, PC, PCBD1, PCDH11X, PCDH19, PCLO, PCNT, PDE6D, PDHA1, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGAP2, PGAP3, PGK1, PGM1, PHC1, PHF6, PHF8, PHGDH, PI4KA, PIBF1, PIGA, PIGB, PIGC, PIGG, PIGH, PIGL, PIGM, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGV, PIGW, PIGY, PIK3CA, PIK3R2, PLA2G6, PLAA, PLEKHG2, PLK4, PLP1, PMM2, PMPCB, PNKP, POGZ, POLG, POLR1C, POLR3A, POLR3B, POLR3K, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PORCN, PPP1CB, PPP1R15B, PPP2CA, PPP2R1A, PPP2R5B, PPP2R5C, PPP2R5D, PPT1, PQBP1, PRPS1, PRSS12, PRUNE1, PSAP, PSAT1, PTCH1, PTCH2, PTCHD1, PTEN, PTPN11, PTS, PURA, PUS3, PYCR2, QARS, QDPR, RAB11B, RAB18, RAB39B, RAB3GAP1, RAB3GAP2, RAB40AL, RAC1, RAC3, RAD21, RAF1, RAI1, RARS, RARS2, RASA2, RBBP8, RBM10, RBMX, RELN, RFT1, RHEB, RIN2, RIT1, RLIM, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF125, RNF135, ROR2, RPGRIP1L, RPL10, RPL11, RPL15, RPL18, RPL26, RPL27, RPL35, RPL35A, RPL5, RPS10, RPS15A, RPS17, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RPS6KA3, RPS7, RRAS, RSRC1, RUSC2, RTTN, RXYLT1, SAMHD1, SASS6, SATB2, SCN1A, SCN2A, SCN8A, SCN9A, SCP2, SDHA, SDHAF1, SEPSECS, SET, SETBP1, SETD2, SETD5, SGSH, SHANK2, SHANK3, SHH, SHOC2, SHROOM4, SIK1, SIX3, SLC12A5, SLC13A5, SLC16A2, SLC17A5, SLC18A2, SLC1A4, SLC25A12,



SLC25A19, SLC25A22, SLC25A46, SLC25A5, SLC33A1, SLC35A1, SLC35A2, SLC35C1, SLC39A8, SLC6A17, SLC6A3, SLC6A8, SLC9A6, SLC9A7, SMARCA2, SMARCA4, SMARCB1, SMARCC2, SMARCE1, SMC1A, SMC3, SMPD1, SMS, SNORD118, SNX14, SON, SOS1, SOS2, SOX10, SOX11, SOX3, SOX4, SOX5, SPG11, SPR, SPTAN1, SRCAP, SRD5A3, SRPX2, SSR4, ST3GAL3, STAG1, STAG2, STAMPB, STIL, STN1, STRADA, STT3A, STT3B, STXBP1, SUFU, SUMF1, SURF1, SYN1, SYNGAP1, SYP, SZT2, TACO1, TAF1, TAF13, TAF2, TARS2, TBC1D20, TBC1D23, TBC1D24, TBC1D7, TBCD, TBCK, TBL1XR1, TBR1, TCF4, TCTN1, TCTN2, TCTN3, TDGF1, TECR, TGFB1, TGIF1, TH, THOC2, TIMM8A, TLK2, TM4SF20, TMC01, TMEM106B, TMEM107, TMEM138, TMEM165, TMEM199, TMEM216, TMEM231, TMEM237, TMEM67, TMLHE, TMTC3, TNIK, TOE1, TOP3A, TPH1, TPH2, TPP1, TRAI, TRAPPC9, TRAPPC12, TRAPPC6B, TRAPPC9, TREM2, TREX1, TRIO, TRIP12, TRMT1, TRMT10A, TRRAP, TSC1, TSC2, TSEN15, TSEN2, TSEN34, TSEN54, TSPAN7, TSR2, TTI2, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBG1, TUBGCP4, TUBGCP6, TUFM, TUSC3, TYMP, TYROBP, UBE2A, UBE3A, UBTF, UFC1, UFM1, UPF3B, USP27X, USP9X, VARS, VARS2, VLDLR, VPS11, VPS13B, VPS33A, VPS53, VRK1, WAC, WARS2, WASHC5, WDFY3, WDR45, WDR45B, WDR62, WDR81, WNT5A, WWOX, ZBTB11, ZBTB18, ZC3H14, ZC4H2, ZCCHC12, ZDHHC15, ZDHHC9, ZEB2, ZFYVE26, ZIC2, ZMYM3, ZMYND11, ZNF335, ZNF41, ZNF423, ZNF674, ZNF711, ZNF81, ZNHIT3

- Klinische Exom Analyse** (nach Absprache)
- Exom Analyse** (nach Absprache)
- Trio-Exom Analyse** (nach Absprache)
- Weitere Panels nach Absprache**