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# Genpanel Heterozygoten-Screening

## Genpanel 'Kingsmore'

**449 Gene**: AAAS, ABCA12, ABCB11, ABCD1, ACAD9, ACADL, ACADM, ACADS, ACADSB, ACADVL, ACE, ACOX1, ADA, ADAMTS13, ADAMTS2, ADAMTSL2, AGA, AGL, AGPS, AGT, AGTR1, AGXT, AHI1, AIRE, ALDH3A2, ALDH5A1, ALDOB, ALG1, ALG6, ALMS1, ALPL, ALS2, AMACR, AMPD1, AMT, ANTXR2, APTX, AR, ARSA, ARSB, ARSE, ARX, ASL, ASPA, ASS1, ATIC, ATM, ATP6V0A2, ATP7A, ATP7B, ATP8B1, ATR, ATRX, AUH, B4GALT1, BCKDHA, BCKDHB, BCS1L, BLM, BMPR2, BTD, BTK, TWNK, C10ORF2, CA2, COQ8A, CBS, CD40LG, CD96, CD96, CDH23, CDKL5, CEP290, CFP, CFTR, CHM, CHRNA1, CHRND, CHRNG, CHRNG, CLDN1, CLDN19, CLN3, CLN5, CLN6, CLN8, CLN8, CLRN1, CNGB3, COL11A2, COL17A1, COL1A2, COL2A1, COL4A3, COL4A4, COL7A1, COQ2, CPS1, CPS1, CPT1A, CPT2, CRLF1, CRTAP, CSTB, CTNS, CTSD, CTSK, CYP11A1, CYP21A2, CYP27A1, CYP27B1, D2HGDH, DCLRE1C, DCX, DDB2, DDC, DGUOK, DHCR24, DHCR7, DKC1, DLD, DLL3, DMD, DMP1, DNAJC19, DNMT3B, DOLK, DPAGT1, DPM1, DPYD, DSP, EDA, EDN3, EDNRB, EFEMP2, EGR2, EIF2AK3, ENPP1, EPM2A, ERBB3, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ESCO2, ETFA, ETFB, ETFDH, ETHE1, F11, F5, FAH, FAM126A, FAM20C, FANCB, FBLN5, FGA, FGB, FGD4, FGFR2, FGG, FH, FKRP, FKTN, FLNA, FOXN1, FOXP3, FRAS1, FREM2, FUCA1, G6PC3, G6PD, GAA, GALC, GALT, GATA1, GBA, GBE1, GCDH, GCSH, GFM1, GJA1, GJB2, GJC2, GLA, GLB1, GLDC, GLE1, GLI3, GNE, GNPTAB, GNS, ADGRV1, GRHPR, GSS, GTF2H5, GUSB, HADH, HADHA, HADHA, HADHB, HAMP, HBA1, HBB, HBB, HESX1, HEXA, HEXB, HFE, HFE2, HGD, HGSNAT, HIBCH, HMGCL, HPRT1, HSD17B10, HSD17B4, HSPG2, HYLS1, IDUA, IGBP1, IGF1, IGHMBP2, ELP1, IKBKG, IL2RG, INSR, INVS, IQCB1, ITGA6, ITGB4, IVD, JAK3, KCNJ1, KCNQ1, KCTD7, KRT18, KRT8, L1CAM, LAMA2, LAMA3, LAMB2, LAMB3, LAMC2, LARGE1, LBR, LEPRE1, LHX3, LIFR, LMNA, LRP2, LRP5, LRPPRC, LYST, MAN2B1, MAPK10, MCCC2, MCOLN1, MECP2, MED12, MEFV, MFSD8, MGAT2, MKS1, MLC1, MMAB, MMACHC, MOCS1, MOCS2, MOGS, MPI, MPL, MPV17, MPZ, MRPS16, MRPS22, MTHFR, MTM1, MTTP, MMUT, MUTYH, MVK, MYO5A, MYO5A, MYO7A, MYO7A, NAGA, NAGS, NBN, NDP, NEB, NEU1, NEUROG3, NHLRC1, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NR0B1, NTRK1, NUP62, OCRL, OFD1, OPA3, OSTM1, OTC, PAH, PAX6, PC, PCDH15, PDHA1, PDHX, PDP1, PDSS1, PDSS2, PEX7, PKHD1, PKLR, PLA2G6, PLCE1, PLEC1, PLEKHG5, PLG, PLOD1, PLP1, PLP1, PMM2, PMP22, PNPO, POLG, POMGNT1, POMT1, POMT2, POU1F1, PPT1, PQBP1, PREPL, PRKAG2, PRKAR1A, PROC, PROP1, PRPS1, PRX, PSAP, PSAT1, PTH1R, RAB23, RAB27A, RAB3GAP1, RAB3GAP2, RAG1, RAG2, RAPSN, RELN, REN, RMRP, RMRP, RPGRIP1L, RS1, SACS, SBDS, SC5D, SCNN1A, SCNN1B, SCNN1G,

SCO2, SELENON, SERPINA1, SFTPA1, SFTPB, SFTPB, SFTPC, SGCA, SGSH, SH2D1A, SIL1, SLC12A1, SLC12A6, SLC16A2, SLC17A5, SLC25A15, SLC25A22, SLC26A2, SLC26A4, SLC34A2, SLC35A1, SLC35C1, SLC35D1, SLC37A4, SLC3A1, SLC4A11, SLC6A8, SLC9A6, SMN1, SMPD1, SNAP29, SOX10, SP110, ST3GAL5, STAR, STRA6, STXBP1, SUCLG1, SUOX, TAZ, TBCE, TCIRG1, TFR2, TGM1, TH, TIMM8A, TK2, TMEM67, TNFRSF11B, TNNT1, TPP1, TREX1, TRIM37, TSEN54, TSFM, TSHB, TSPYL1, TTN, TTPA, UBA1, UBR1, UQCRB, UQCRQ, UROS, USH1C, USH1G, USH2A, VDR, VLDLR, VPS13B, VPS33B, WAS, WNT10A, WNT3, WNT7A, WT1, XPA, ZIC3, ZMPSTE24, ZNF469

**Referenz**: Kingsmore S., Comprehensive Carrier Screening and Molecular Diagnostic Testing for Recessive Childhood Diseases. PLOS Currents Evidence on Genomic Tests, 2012

Es werden nur Varianten der Klasse 4 (wahrscheinlich pathogen) und 5 (pathogen) berichtet (gemäss Richards S. et al., *Genet Med*, 2015).

Die aufgeführten Gene entsprechen den der jeweiligen Referenz zugrundeliegenden Empfehlungen. Die gemäss Genomics England PanelApp aufgeführten Gene entsprechen den als «diagnostic-grade», also diagnostisch gewerteten Genen (s.a. https://panelapp.genomicsengland.co.uk/#!Guidelines).

Die Auswertung weiterer Gene ist nach Rücksprache mit unserem Labor möglich.