

Centobrief January 2017 :: newly added single gene analyses and NGS panels

CENTOGENE has recently added the following single gene tests and Next Generation Sequencing panels to its portfolio. CENTOGENE's NGS panel composition always reflects the clinical relevance to the specific disease phenotype. All of our NGS panels are processed with a standardized quality scheme and internal controls.

Benefit from our medical expertise and first class genetic testing.

New single genes	
Disease	Gene
Metabolic Diseases	
Lactate dehydrogenase-B deficiency	LDHB
Chylomicron retention disease	SAR1B
Neurological Diseases	
Mitochondrial complex IV deficiency	APOPT1
Bone, Skin and Immune Diseases	
Dyschromatosis universalis hereditaria type 3	ABCB6
Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency	GGCX
Epidermolysis bullosa dystrophica, autosomal recessive, modifier of	MMP1
Cardiological and Lung Diseases	
Thoracic aortic aneurysm dissection	SMAD2
Haematological Diseases	
Vitamin K-dependent clotting factors combined deficiency type 1	
Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency	GGCX

New NGS panels or extended NGS panels

Disease	Genes
<p>Neurological Diseases</p> <p>Allneuro panel (updated; new panel composition is displayed)</p>	<p>A2M, AAAS, AANAT, AARS, AARS2, AASS, ABAT, ABCA1, ABCB7, ABCC6, ABCC8, ABCD1, ABCD4, ABHD12, ABHD5, ACACA, ACAD9, ACADL, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACE, ACKR1, ACO2, ACOX1, ACSF3, ACSL4, ACTA1, ACTA2, ACTB, ACTG1, ACVRL1, ACY1, ADAM10, ADAR, ADCK3, ADCY5, ADGRG1, ADGRV1, ADK, ADNP, ADSL, AFF2, AFG3L2, AGA, AGK, AGL, AGRN, AGXT, AHCY, AHI1, AIFM1, AIMP1, AKT1, AKT3, ALAD, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ALMS1, ALOX5AP, ALS2, ALX1, ALX3, ALX4, AMACR, AMN, AMPD1, AMPD2, AMT, ANG, ANK3, ANKRD11, ANO10, ANO3, ANO5, AP1S1, AP1S2, AP3B1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOA1, APOE, APP, APTX, AR, ARFGF2, ARG1, ARHGAP31, ARHGEF10, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARL13B, ARL6, ARSA, ARSB, ARSE, ARSI, ARX, ASAH1, ASCL1, ASL, ASNS, ASPA, CNBP, ASS1, ASXL1, ASXL3, ATCAY, ATIC, ATLL1, ATM, ATP13A2, ATP1A2, ATP1A3, ATP2A1, ATP2A2, ATP2B3, ATP2B4, ATP5E, ATP6AP2, ATP6V0A2, ATP7A, ATP7B, ATP8A2, ATPAF2, ATR, ATRX, ATXN3, B3GLCT, B4GALNT1, B4GALT1, B9D1, B9D2, BAG3, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCKDHA, BCKDHB, BCKDK, BCOR, BCS1L, BDNF, BEST1, BICD2, BIN1, BLOC1S3, BLOC1S6, BOLA3, BRAF, BRAT1, BRWD3, BSLC2, BSNL, C10orf2, C12orf57, C12orf65, C19orf12, C5orf42, C9orf72, CA8, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1H, CACNA1S, CACNB4, CACNG2, CAMTA1, CAPN3, CASC5, CASK, CASR, CAV3, CBL, CC2D1A, CC2D2A, CCDC28B, CCDC78, CCDC88C, CCM2, CCT5, CD207, CD320, CD36, CD59, CD96, CDH15, CDK11A, CDK5RAP2, CDKL5, CDON, CENPJ, CEP135, CEP152, CEP290, CEP41, CEP63, CERS1, CFL2, CHAT, CHCHD10, CHD2, CHD7, CHD8, CHMP1A, CHMP2B, CHRM3, CHRNA1, CHRNA2, CHRNA4, CHRNB1, CHRNB2, CHRND, CHRNE, CHRNG, CHSY1, CISD2, CISH, CI21, CLCN1, CLCN2, CLCNKA, CLCNKB, CLIC2, CLN3, CLN5, CLN6, CLN7, CLN8, CLN9, CLN10, CNTNAP2, CNTNAP4, COA5, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL12A1, COL18A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, COL7A1, COLQ, COQ2, COQ9, COX10, COX15, COX6A1, COX6B1, CP, CPA6, CPT2, CR1, CRADD, CRBN, CREBBP, CRYAB, CSF1R, CSF2RB, CSPP1, CST3, CSTB, CTC1, CTDP1, CTNBB1, CTSD, CUL4B, CWF19L1, CYP11B2, CYP27A1, CYP2U1, CYP7B1, DAG1, DARS2, DBT, DCAF17, DCTN1, DCX, DDHD1, DDHD2, DDOST, DEPDC5, DES, DGUOK, DHCR7, DHH, DHTKD1, DIAPH3, DKC1, DLD, DLG3, DMD, DNAH9, DNAJB2, DNAJB6, DNAJC5, DNAJC6, DNM1L, DNM2, DNMT1, DOCK8, DOK7, DOLK, DPAGT1, DPM1, DPM3, DRD2, DRD3, DSC3, DST, DTNBP1, DYNC1H1, DYNC2H1, DYRK1A, DYSF, EARS2, EBP, EDN3, EDNRB, EEF2, EFHC1, EFTUD2, EGR2, EHMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF4E, EIF4G1, ELK1, ELOVL4, ELOVL5, EMD, EMX2, ENO3, ENTPD1, EOMES, EP300, EPB41L1, EPM2A, ERBB4, ERCC1, ERCC2, ERCC5, ERCC6, ERLIN1, ERLIN2, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EXOC8, EXOSC3, EXOSC8, F2, F5, FA2H, FADD, FAM126A, FAM134B, FANCB, FASTKD2, FBLN5, FBXO38, FBXO7, FCGR2B, FGA, FGD1, FGD4, FGF10, FGF14, FGFR1, FGFR2, FGFR3, FH, FHL1, FIG4, FKRP, FKTN, FLNA, FLNC, FLRT1, FLVCR1, FLVCR2, FMR1, FOLR1, FOXP1, FOXP2, FOXP3, FOXRED1, FREM1, FRG1, FRMD7, FTL, FTO, FTSJ1, FUS, FXN, FZD9, GAA, GABRA1, GABRB3, GABRD, GABRG2, GAD1, GALT, GALS, GAMT, GAN, GAN, GARS, GATM, GBA, GBA2, GBE1, GCDH, GCH1, GCSH, GDAP1, GDI1, GDNF, GFAP, GFER, GFM1, GFPT1, GIGYF2, GJB1, GJB3, GJC2, GK, GLB1, GLDC, GLI2, GLI3, GLRA1, GLRB, GM2A, GMPBB, GNAL, GNB4, GNE, GNPAT, GNS, GOSR2, GP1BA, GPC3, GPR143, GRIA3, GRID2, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIP1, GRM1, GRN, GSN, GUSB, GYG1, GYS1, HADHA, HADHB, HCCS, HDAC4, HDAC8, HEPACAM, HERC2, HESX1, HEXA, HEXB, HFE, HGSNAT, HINT1, HK1, HMGCS2, HNRNPDL, HOXA1, HOXD10, HPCA, HPD, HPRT1, HPS1, HPS3, HPS4, HPS5, HPS6, HPSE2, HRAS, HSD17B10, HSD17B4, HSPB1, HSPB3, HSPB8, HSPD1, HTRA1, HTRA2, HTT, HUWE1, HYAL1, ICAM1, ICK, IDS, IDUA, IER3IP1, IFRD1, IFT140, IFT27, IGBP1, IGF1, IGHMBP2, IKBKAP, IKBKG, IL11RA, IL1RAPL1, IL1RN, INF2, INPP5E, INS, IQSEC2, IRX5, ISPD, ITM2B, ITPR1, JRK, KANK1, KARS, KAT6B, KBTBD13, KCNA1, KCNC3, KCND3, KCNE1L, KCNJ1, KCNJ10, KCNJ11, KCNK18, KCNK9, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCCTD7, KDM5C, KDM6A, KIAA0196, KIAA2022, KIF11, KIF1A, KIF1B, KIF1C, KIF21A, KIF5A, KIF7, KIRREL3, KLF8, KMT2A, KMT2D, KRAS, KRIT1, KRT5, L1CAM, LAMA2, LAMB1, LAMB2, LAMC3, LAMP2, LARGE, LBR, LDB3, LDHA, LGI1, LHX4, LIMS2, LINS, LITAF, LMBRD1, LMNA, LMNB1, LPIN1, LPIN2, LRP2, LRPPRC, LRRK2, LRSAM1, LYZ, LZTFL1, MAG, MAGEL2, MAGI2, MAGT1, MAMLD1, MAN1B1, MAOA, MAPK10, MAPT, MARS, MARS2, MASP1, MATR3, MBD5, MBTPS2, MCEE, MCPH1, MECP2, MED12, MED17, MED23, MED25, MEF2C, MET, MFN2, MFSDB, MGAT2, MID1, MKKS, MKS1, MLC1, MMAA, MMAB, MMACHC, MMADHC, MOGS, MPDU1, MPDZ, MPI, MPV17, MPZ, MRE11A, MSX1, MSX2, MTFMT, MTHFR, MTM1, MTFMR14, MTFMR2, MTO1, MTOR, MTPAP, MTR, MTRR, MUSK, MUT, MVK, MYCN, MYF6, MYH14, MYH7, MYH9, MYO5A, MYOT, NAA10, NAGA, NAGLU, NAT8L, NBN, NDE1, NDP, NDRG1, NDST1, NDUFA1, NDUFA10, NDUFA12, NDUFA2, NDUFA2, NDUFA9, NDUFA9, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEB, NEDD4L, NEFH, NEFL, NEU1, NF1, NFIX, NFU1, NGF, NHEJ1, NHLRC1, NHS, NIPA1, NIPBL, NKX2-1, NLGN3, NLGN4X, NLRP12, NLRP3, NOD2, NOG, NOL3, NPS56, NOS2, NOS3, NOTCH2, NOTCH3, NPC1, NPC2, NPHP1, NPHP3, NRAS, NRG1, NRXN1, NSD1, NSDHL, NSUN2, NT5C2, NTRK1, NTRK2, NUBPL, NXF5, OCLN, OCRL, OFD1, OPA1, OPA3, OPHN1, OPTN, ORC1, OTC, PAFAH1B1, PAH, PAK3, PANK2, PARK2, PARK7, PAX6, PC, PCBD1, PCDH19, PCK2, PCNT, PDCD10, PDE6D, PDE8B, PDHA1, PDK3, PDS1, PDS2, PDYN, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PFN1, PGAM2, PGAP1, PGK1, PGM1, PHF6, PHF8, PHKA1, PHOX2B, PHYH, PIEZO2, PIGA, PIGL, PIGN, PIGO, PIGV, PIK3CA, PIK3R2, PIK3R5, PINK1, PLA2G6, PLCB1, PLEC, PLEKHG4, PLEKHG5, PLP1, PMM2, PMP22, PNKD, PNKP, PNPLA2, PNPLA6, PNPO, POGZ, POLG, POLG2, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, PONT1, POMT2, PORCN, PPT1, PQBP1, PREPL, PRICKLE1, PRICKLE2, PRKAG2, PRKCG, PRKCH, PRKRA, PRNP, PROP1, PRPH2, PRPS1, PRRT2, PRRX1, PRSS12, PRX, PSAP, PSEN1, PSEN2, PTCH1, PTEN, PTF1A, PTPN11, PTS, PUS1, PVRL1, PYGM, QARS, QDPR, RAB18, RAB39B, RAB3GAP1, RAB3GAP2, RAB40AL, RAB7A, RABGGTA, RAD21, RAD50, RAF1, RAI1, RANBP2, RAPS, RARS2, RBBP8, RBFOX1, RBFOX3, RBM10, RBM8A, REEP1, REEP2, RELN, RET, RFT1, RIN2, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF135, RNF170, RNU4ATAC, ROGDI, ROR2, RRPGRIP1L, RPL10, RPS6KA3, RRM2B, RTN2, RUBCN, RYR1, SACS, SAMHD1, SBF1, SBF2, SCARB2, SCN10A, SCN1A, SCN1B, SCN2A, SCN3A, SCN4A, SCN5A, SCN8A, SCN9A, SCO1, SCO2, SCP2, SDCCAG8, SDHA, SDHAF1, SEPN1, SEPSECS, SERPINI1, SETBP1, SETD2, SETX, SGCA, SGCB, SGCD, SGCE, SGCG, SGSH, SH3TC2, SHANK2, SHANK3, SHH, SHOC2, SHROOM4, SIGMAR1, SIL1, SIX3, SIX6, SKI, SLC12A6, SLC13A5, SLC16A2, SLC17A5, SLC19A3, SLC1A3, SLC20A2, SLC22A5, SLC25A12, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC25A4, SLC2A1, SLC30A10, SLC33A1, SLC35A1, SLC35A2, SLC35C1, SLC3A1, SLC4A1, SLC4A4, SLC5A7, SLC6A1, SLC6A3, SLC6A5, SLC6A8, SLC9A6, SLC9A9, SMARCA2, SMARCA4, SMARCB1, SMC1A, SMC3, SMCHD1, SMN1, SMN2, SMPD1, SMS, SNAP29, SNCA, SNCAIP, SNCB, SNIP1, SNX3, SOBP, SOD1, SORL1, SOS1, SOX10, SOX3, SPAST, SPG11, SPG20, SPG21, SPG7, SPR, SPTAN1, SPTBN2, SPTLC1, SPTLC2, SRD5A3, SRPX2, ST3GAL3, ST3GAL5, STIL, STRADA, STUB1, STXBP1, SUCLA2, SUCLG1, SUMF1, SURF1, SYN1, SYNE1, SYNE2, SYNGAP1, SYNJ1, SYP, SYT14, SZT2, TACO1, TAF1, TAF2, TARDBP, TAS2R38, TAZ, TBC1D24, TBCE, TBL1XR1, TBX1, TCAP, TCF4, TCTN1, TCTN2, TCTN3, TDP1, TECPR2, TECR, TFAP2A, TFAP2B, TFG, TG, TGFBI, TGFBI2, TGFBR1, TGFBR2, TGIF1, TGM6, TH, THAP1, TICAM1, TIMM8A, TIRAP, TK2, TLR3, TLR5, TMO1, TMEM126A, TMEM138, TMEM165, TMEM216, TMEM231, TMEM237, TMEM67, TMEM70, TMLHE, TNF, TNFSF4, TNNT1, TNPO3, TOR1A, TP63, TPK1, TPM2, TPM3, TPP1, TRAF3, TRAPPC11, TRAPPC9, TREM2, TREX1, TRIM2, TRIM32, TRPM6, TRPM7, TRPS1, TRPV4, TSC1, TSC2, TSEN2, TSEN34, TSEN54, TSFM, TSHB, TSHR, TSPAN7, TTBK2, TTC19, TTC21B, TTC8, TTI2, TTN, TTPA, TTR, TUBA1A, TUBA4A, TUBA8, TUBB2B, TUBB3, TUBB4A, TUBGCP6, TUSC3, TWIST1, TYMP, TYROBP, UBA1, UBE2A, UBE3A, UBQLN2, UCHL1, UMPS, UNC93B1, UPB1, UPF3B, UQCRCB, UQCRCQ, USP8, USP9X, VAMP1, VANGL1, VAPB, VAX1, VCP, VDAC1, VEGFA, VHL, VIPAS39, VLDLR, VPS13A, VPS13B, VPS35, VPS37A, VRK1, WAC, WDCPC, WDR45, WDR48, WDR62, WDR81, WFS1, WNK1, WNT10A, WNT3, WNT5A, WNT7A, WWOX, XBP1, XK, YAP1, YARS, YWHAE, ZBTB16, ZBTB18, ZCCHC12, ZDHHC15, ZDHHC9, ZEB2, ZFR, ZFYVE26, ZFYVE27, ZIC2, ZIC3, ZNF335, ZNF423, ZNF592, ZNF711, ZNF81</p>

New NGS panels or extended NGS panels	
Disease	Genes
Neurological Diseases	
Non-dystrophic myotonia congenita panel (new)	ATP2A1, CACNA1A, CACNA1S, CAV3, CLCN1, HINT1, HSPG2, KCNA1, KCNE3, SCN4A
Ophthalmological Diseases	
Cataract panel (updated)	AGK, BCOR , BFSP1 , BFSP2 , CHMP4B , CRYAA, CRYAB, CRYBA1 , CRYBA2 , CRYBA4 , CRYBB1, CRYBB2 , CRYBB3, CRYGB , CRYGC , CRYGD , CRYGS , CTDP1, EPHA2 , EYA1 , FOXC1 , FOXE3 , FTL , FYCO1, GALK1 , GCNT2, GJA3 , GJA8, HSF4, LEMD2 , LIM2, LSS, MAF, MIP, NHS, P3H2, PAX6, PITX3, SIPA1L3, SLC16A12, TDRD7, UNC45B , VIM , VSX2 , WFS1
Bone, Skin and Immune Diseases	
Epidermolysis bullosa panel (extended)	CHST8 , COL17A1, COL7A1 , CSTA , DSG1 , DSP, DST, EXPH5, FERMT1, ITGA3, ITGA6, ITGB4, JUP, KRT14, KRT5, LAMA3, LAMB3, LAMC2, MMP1 , PKP1, PLEC, TGM5
Cardiological and Lung Diseases	
Comprehensive pulmonary disease panel (new)	ABCA3, ACVRL1, AP3B1, ASCL1, BDNF, BLOC1S3, BLOC1S6, BMPR2, CCDC39, CCDC40, CFTR, CSF2RA, CSF2RB, DKC1, DNAAF1, DNAAF2, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DOCK8, DTNBP1, EDN3, EFEMP2, ELMOD2, ELN, ENG, FBLN5, FBN1, FLCN, FOXF1, GDNF, HPS1, HPS3, HPS4, HPS5, HPS6, LTBP4, MUC5B, NKX2-1, NME8, NOP10, PARN, PHOX2B, RET, RSPH1, RSPH4A, RSPH9, RTEL1, SCNN1A, SCNN1B, SCNN1G, SERPINA1, SFTPA1, SFTPA2, SFTPB, SFTPC, SFTPD, SMAD9, STAT3, TERC, TERT, TINF2, TSC1, TSC2
Vascular Diseases	
Aortic aneurysm, hereditary thoracic panel (old name) Familial thoracic aortic aneurysm panel (new name) (updated)	ACTA2, BGN , CBS, LOX , MAT2A , MFAP5 , MYH11, MYLK
Malformation and Retardation Syndromes	
Marfan, Loews-Dietz syndrome and related disorders panel (new)	COL3A1, COL5A1, COL5A2, EFEMP2, FBN1, FBN2, NOTCH1, SKI, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFB3, TGFB3, TGFB3
Micro syndrome panel (extended)	ALDH18A1 , CREBBP, CUL7, RAB18, RAB3GAP1, RAB3GAP2, TBC1D20