ASPER
NEUROGENETICS

Asper Neurogenetics

Disease/Condition name	Method	No of detectable genes/markers	Genes	Price (EUR)
Alzheimer Disease	Sanger Sequencing	1/2	APOE	87
Amyotrophic Lateral Sclerosis	NGS	22	ALS2, ANG, CHCHD10, CHMP2B, ERBB4, FIG4, FUS, MATR3, OPTN, PFN1, SETX, SIGMAR1, SOD1, SPART, SPG11, SQSTM1, TARDBP, TBK1, TUBA4A, UBQLN2, VAPB, VCP	1070
Autism Spectrum Disorders	NGS	62	ADNP, ADSL, ANKRD11, ARX, AVPR1A, BRAF, CACNA1C, CDKL5, CHD2, CHD7, CNTNAP2, CREBBP, DHCR7, EHMT1, EN2, FOXG1, FOXP1, FOXP2, GABRB3, HDAC8, HOXA1, HPRT1, MAGEL2, MECP2, MED12, MID1, NHS, NIPBL, NLGN3, NLGN4X, NRXN1, NSD1, NTNG1, OXTR, PCDH19, PDE8B, POGZ, PTCHD1, PTEN, PTPN11, PQBP1, RAD21, RAI1, RELN, RPL10, SCN1A, SCN2A, SETD2, SHANK2, SHANK3, SLC6A4, SLC6A8, SLC9A9, SMC3, SMC1A, TBL1XR1, TCF4, TSC1, TSC2, UBE3A, VPS13B, ZEB2	1338
Charcot-Marie-Tooth Disease	NGS	67	AARS, AIFM1, ARSA, BSCL2, C12orf65, COX6A1, DCTN1, DHTKD1, DNAJB2, DNM2, DYNC1H1, EGR2, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, GNB4, HARS, HINT1, HK1, HSPB1, HSPB3, HSPB8, IGHMBP2, INF2, KARS, KIF1A, KIF5A, KIF1B, LITAF, LMNA, LRSAM1, MARS, MED25, MFN2, MORC2, MPZ, MTMR2, NAGLU, NDRG1, NGF, PDK3, PLEKHG5, PMP22, POLG, PRPS1, PRX, RAB7A, REEP1, SBF1, SBF2, SCN9A, SETX, SH3TC2, SLC5A7, SPTLC1, SPTLC2, SURF1, TFG, TRIM2, TRPV4, TYMP, VCP, WNK1, YARS	1314
	MLPA	2	KIF1B, PMP22	310
	MLPA	1	GJB1	310
	MLPA	2	MFN2, MPZ	310
	MLPA	5	GARS, HSPB1, HSBP8, RAB7A, SPTLC1	590
	MLPA	7	EGR2, GDAP1, MTMR2, NEFL, SBF2, SH3TC2, PRX	590
Congenital Myasthenic Syndrome	NGS	23	AGRN, ALG2, ALG14, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COLQ, DOK7, DPAGT1, GFPT1, GMPPB, LAMB2, LRP4, MUSK, PLEC, PREPL, RAPSN, SCN4A, SLC5A7, SNAP25, SYT2	1070
Congenital Myopathy and Distal Myopathy	NGS	42	ACTA1, ANO5, BAG3, BIN1, CAV3, CCDC78, CFL2, CNTN1, COL6A1, COL6A3, COL12A1, CRYAB, DES, DNAJB6, DNM2, DYSF, FHL1, FLNC, GNE, KLHL40, KLHL41, LDB3, LMOD3, MATR3, MEGF10, MICU1, MTM1, MTMR14, MYF6, MYH7, MYOT, NEB, RYR1, SELENON, STAC3, SQSTM1, TIA1, TNNT1, TPM2, TPM3, TTN, VCP	1338
	MLPA	2	MTM1, MTMR1	590
Cornelia de Lange Syndrome	NGS	9	AFF4, ANKRD11, HDAC8, KMT2A, NIPBL, RAD21, SMC3, SMC1A, TAF6	1030
	MLPA	1	NIPBL	590

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Craniosynostosis	NGS, Sanger Sequencing	7	AFF4, ANKRD11, HDAC8, KMT2A, NIPBL, RAD21, SMC3, SMC1A, TAF6	1051
	MLPA	1	TWIST1	310
Dystonia	NGS	39	ACTB, ADCY5, ANO3, ARSA, ATM, ATP1A3, ATP7B, CACNA1B, CIZ1, COL6A3, DRD2, GCDH, GCH1, GNAL, GNAO1, HPCA, KCNMA1, KCTD17, PANK2, PLA2G6, PNKD, PRKN, PRKRA, PRRT2, RELN, SGCE, SLC2A1, SLC6A3, SLC25A1, SLC30A10, SLC39A14, SPR, TAF1, TBCE, TH, THAP1, TIMM8A, TOR1A, TUBB4A	1314
	MLPA	7	ATP1A3, GCH1, PRKRA, SGCE, TH, THAP1, TOR1A	710
Epilepsy	NGS	184	AARS, ABAT, ACY1, ADAR, ADSL, ALDH5A1, ALDH7A1, ALG3, ALG13, ARHGEF9, ARHGEF15, ARX, ASAH1, ATP1A2, ATP1A3, ATP6AP2, ATRX, BRAT1, CACNA1A, CACNA1D, CACNA2D2, CACNA1H, CACNB4, CASK, CDC42, CDKL5, CERS1, CHD2, CHRNA2, CHRNA4, CHRNA7, CHRNB2, CLCN2, CLN3, CLN8, CNTN2, CNTNAP2, C12orf57, CPA6, CRH, CSTB, CTSF, DEPDC5, DHFR, DNAJC5, DNM1, DNM1L, DOCK7, DYRK1A, EEF1A2, EFHC1, EPM2A, FGF12, FLNA, FOLR1, FOXG1, GABBR2, GABRA1, GABRB1, GABRB3, GABRD, GABRG2, GAMT, GATM, GLDC, GNAO1, GOSR2, GPHN, GRIN1, GRIN2A, GRIN2B, GRIN2D, HCN1, HNRNPU, HUWE1, IER3IP1, ITPA, IQSEC2, KANSL1, KCNA1, KCNA2, KCNB1, KCNC1, KCNH1, KCNJ10, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCTD7, KIF1A, KIF5C, LGI1, LIAS, MBD5, MCCC1, MDH2, MECP2, MEF2C, MFSD8, MOCS1, MOCS2, MTOR, NACC1, NECAP1, NEXMIF, NGLY1, NHLRC1, NOL3, NPRL2, NR2F1, NRXN1, PCDH19, PIK3R2, PIGA, PIGN, PIGO, PIGT, PLCB1, PLPBP, PNKP, PNPO, POLG, PPT1, PRDM8, PRICKLE1, PRICKLE2, PRRT2, PURA, QARS, RBFOX1, RBFOX3, RELN, ROGDI, SATB2, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SERPINI1, SIK1, SLC1A2, SLC12A5, SLC13A5, SLC19A3, SLC25A22, SLC2A1, SLC35A2, SLC35A3, SLC6A1, SLC6A8, SLC9A6, SMARCA2, SMC1A, SNIP1, SNX27, SPATA5, SPTAN1, SRPX2, ST3GAL3, ST3GAL5, STX1B, STXBP1, SYN1, SYNGAP1, SYNJ1, SYP, SZT2, TBC1D24, TBCK, TCF4, TPP1, TSC1, TSC2, TUBB3, UBA5, UBE3A, WDR45, WWOX, ZDHHC9, ZEB2	1546
	MLPA	11	CHRNA4, CHRNB2, EPM2A, KCNQ1, KCNQ3, NHLRC1, PCDH19, SCN1A, SLC2A1, STXBP1, WWOX	1334
Fragile X Syndrome	Repeat Expansion/ Fragment Length Analysis	1	FMR1	262

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Frontotemporal Dementia	NGS	5	CHMP2B, GRN, MAPT, TARDBP, PSEN1	1051
	MLPA	3	CRHR1, GRN, MAPT	590
Hereditary Spastic Paraplegia	NGS	34	ATL1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, B4GALNT1, BSCL2, CYP7B1, CYP2U1, DDHD2, ERLIN2, FA2H, GBA2, GJC2, HSPD1, KIF1A, KIF5A, L1CAM, NIPA1, PLP1, PNPLA6, REEP1, RTN2, SLC16A2, SPART, SPAST, SPG7, SPG11, SPG21, TECPR2, VPS37A, WASHC5, ZFYVE26	1314
	Sanger Sequencing	1/1	MT-ATP6	87
	MLPA	5	ATL1, NIPA1, SPAST, SPG7, REEP1	806
Joubert Syndrome	NGS	29	AHI1, ARL13B, B9D1, B9D2, C5orf42, CC2D2A, CEP290, CEP41, CEP104, CSPP1, INPP5E, KIF7, KIAA0556, KIAA0586, MKS1, NPHP1, OFD1, PDE6D, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM67, TMEM237, TTC21B, ZNF423	1314
Leukodystrophy and Leukoencephalopathy	NGS, Sanger Sequencing	40	ABCD1, ADAR, AIMP1, ARSA, ASPA, CLCN2, CSF1R, DARS2, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, FAM126A, FOLR1, GALC, GFAP, GJC2, HEPACAM, HSPD1, HTRA1, L2HGDH, LMNB1, MLC1, NOTCH3, PLP1, POLR3A, POLR3B, PSAP, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, SAMHD1, SCP2, SOX10, SUMF1, TREX1, TUBB4A	1070
	MLPA	7	ABCD1, ASPA, L2HGDH, LMNB1, MLC1, NOTCH3, PLP1	1070
Limb-Girdle Muscular Dystrophy	NGS	27	ANO5, CAPN3, CAV3, DAG1, DES, DNAJB6, DYSF, FKTN, GMPPB, HNRNPDL, ISPD, LMNA, MYOT, PLEC, POMGNT1, POMK, POMT1, POMT2, SGCA, SGCB, SGCD, SGCG, TCAP, TNPO3, TRAPPC11, TRIM32, TTN	1338
	MLPA	12	ANO5, CAPN3, DYSF, FKRP, LCAV3, MNA, MYOT, SGCA, SGCB, SGCD, SGCG, ZMPSTE24	1343
Menkes Disease	Sanger Sequencing	1	ATP7A	1030
	MLPA	1	ATP7A	310
Microcephaly	NGS	24	AP4M1, ASPM, CASC5, CASK, CDK5RAP2, CENPJ, CEP63, CEP135, CEP152, EFTUD2, IER3IP1, KIF11, MCPH1, NDE1, NHEJ1, PAFAH1B1, PCNT, PNKP, POMT1, SLC25A19, STIL, TUBB2B, TUBGCP6, WDR62	1314
Mitochondrial Diseases	NGS	37	Mitochondrial Genome	803

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Mitochondrial Diseases	NGS	133	AARS2, ABCB7, ACAD9, ACADL, ACADM, ACADS, ACADVL, AFG3L2, AIFM1, ALAS2, APTX, ATPAF2, ATP5F1E, AUH, BCS1L, BOLA3, C12orf65, CISD2, COA5, COQ2, COQ6, COQ9, COQ8A, COX10, COX15, COX6B1, CPT1A, CPT2, DARS2, DGUOK, DLAT, DLD, DNAJC19, DNM1L, ETFA, ETFB, ETFDH, ETHE1, FASTKD2, FBP1, FH, FOXRED1, G6PC, GAMT, GATM, GFER, GFM1, GYS2, HARS2, HLCS, HADH, HADHA, HSPD1, ISCU, LRPPRC, MFN2, MPV17, MRPS16, MRPS22, MTFMT, MTPAP, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NUBPL, OPA1, OPA3, PC, PDHA1, PDHB, PDHX-,PDP1, PDSS1, PDSS2, PDX1, POLG, POLG2, PUS1, RARS2, REEP1, RRM2B, SARS2, SCO1, SCO2, SDHA, SDHAF1, SETX, SLC19A3, SLC25A20, SLC25A3, SLC25A4, SLC6A8, SLC37A4, SOD1, SPG7, SUCLA2, SUCLG1, SURF1,TACO1, TAZ, TIMM8A, TK2, TMEM126A, TMEM70, TRMU, TSFM, TTC19, TUFM, TWNK, TYMP, UQCRB, UQCRQ, WFS1, YARS2	1314
	MLPA	10	DGUOK, MPV17, POLG, POLG2, RRM2B, SLC25A4, SUCLA2, SUCLG1, TK2, TWNK	710
Mitochondrial Encephalomyopathy Lactic Acidosis and Stroke-Like Episodes (MELAS Syndrome)	Sanger Sequencing	1/1	MT-TL1	87
Neurodegeneration with Brain Iron Accumulation	NGS	10	ATP13A2, COASY, C19orf12, CP, DCAF17, FA2H, FTL, PANK2, PLA2G6, WDR45	1051
	MLPA	2	PANK2, PLA2G6	590
Parkinson's Disease	NGS	32	ADH1C, ATP1A3, ATP13A2, ATP6AP2, ATXN2, DCTN1, DNAJC6, EIF4G1, FBXO7, FTL, GBA, GCH1, GIGYF2, HTRA2, LRRK2, MAPT, PARK7, PINK1, PLA2G6, PRKN, PRKRA, SLC6A3, SLC30A10, SNCA, SNCB, SPR, SYNJ1, TAF1, TBP (excluding exon 3), TH, UCHL1, VPS35	1314
	MLPA	7	GCH1, LRRK2, PARK7, PINK1, PRKN, SNCA, UCHL1	710
Short Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency	Sanger Sequencing	1	ACADS	515
Smith-Lemli-Opitz Syndrome	Sanger Sequencing	1	DHCR7	525

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Spinocerebellar Ataxias	NGS	66	ABCB7, ABHD12, ACO2, AFG3L2, ANO10, APTX, ATCAY, ATM, ATP8A2, CASK, CCDC88C, CLCN2, CLN5, COQ8A, CWF19L1, CYP27A1, DARS2, FXN, GOSR2, GRM1, DNMT1, EEF2, ELOVL4, ELOVL5, FGF14, FLVCR1, ITPR1, KCNC3, KCND3, KCNJ10, LAMA1, NOP56, NPC1, NPC2, OPHN1, PDYN, PHYH, PNKP, PNPLA6, POLG, PRKCG, RUBCN, PTF1A, RNF216, SACS, SETX, SIL1, SLC9A1, SLC9A6, SLC52A2, SNX14, SPTBN2, STUB1, SYNE1, SYT14, TTBK2, TDP1, TGM6, TMEM240, TPP1, TTPA, TUBB4A, TWNK, WFS1, WWOX, ZNF592	1314
	Repeat Expansion Analysis	13	ATXN1, ATXN2, ATXN3, ATXN7, ATXN8, ATXN10, ATN1, BEAN1, CACNA1A, FXN, NOP56, PPP2R2B, TBP	3480
Tuberous Sclerosis	NGS	2	TSC1, TSC2	803
	MLPA	2	TSC1, TSC2	590
Very Long Chain Acyl-Coenzyme A Dehydrogenase (VLCAD) Deficiency	NGS	1	ACADVL	803
	MLPA	1	ACADVL	310
Wilson Disease	Sanger Sequencing	1	ATP7B	773
	MLPA	1	ATP7B	310
Whole Exome Sequencing (WES)	NGS		Solo sample Trio samples	1567 3100
Whole Genome Sequencing (WGS)	NGS		Solo sample Trio samples	4500 8800

Service includes
- DNA extraction
- Genotyping
- Bioinformatic analysis of detected variants
- Detected variants available in Excel format (on request)
- Validation of phenotype/diagnosis associated known or possibly pathogenic variants
- Clinical and biological interpretation
- Results delivery by registered mail (on request)
- TAT 2-9 weeks, excl. WES and WGS 15 weeks

Online ordering, specimen requirements and shipping instructions are available at www.asperbio.com.

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