ASPER
NEUROGENETICS

Asper Neurogenetics

Disease/Condition name	No of detectable genes/markers	Genes	Method	Price (EUR)
Alzheimer Disease	1/2	APOE	Sanger Sequencing	87
Amyotrophic Lateral Sclerosis	22	ALS2, ANG, CHCHD10, CHMP2B, ERBB4, FIG4, FUS, MATR3, OPTN, PFN1, SETX, SIGMAR1, SOD1, SPART, SPG11, SQSTM1, TARDBP, TBK1, TUBA4A, UBQLN2, VAPB, VCP	NGS NGS with CNV	1030 1400
Autism Spectrum Disorders	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	NGS NGS with CNV	1030 1400
Brunner Syndrome	1	MAOA	NGS NGS with CNV	843 1400
Charcot-Marie-Tooth Disease	75	AARS, ABHD12, AIFM1, ARHGEF10, ARSA, ATP1A1, BSCL2, C12orf65, COX6A1, DCTN1, DHTKD1, DNAJB2, DNM2, DNMT1, 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, MCM3AP, MED25, MFN2, MME, MORC2, MPZ, MTMR2, NAGLU, NDRG1, NEFH, NGF, PDK3, PLEKHG5, PMP22, POLG, PRPS1, PRX, RAB7A, REEP1, SBF1, SBF2, SCN9A, SETX, SH3TC2, SLC5A7, SPTLC1, SPTLC2, SURF1, TFG, TRIM2, TRPV4, TYMP, VCP, WARS, WNK1, YARS	NGS NGS with CNV	1300 1700
	2	KIF1B, PMP22	MLPA	310
	1	GJB1	MLPA	310
	2	MFN2, MPZ	MLPA	310
	5	GARS, HSPB1, HSBP8, RAB7A, SPTLC1	MLPA	590
	7	EGR2, GDAP1, MTMR2, NEFL, SBF2, SH3TC2, PRX	MLPA	590
Congenital Myasthenic Syndrome	23	AGRN, ALG2, ALG14, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COLQ, DOK7, DPAGT1, GFPT1, GMPPB, LAMB2, LRP4, MUSK, PLEC, PREPL, RAPSN, SCN4A, SLC5A7, SNAP25, SYT2	NGS NGS with CNV	1030 1400
Congenital Myopathy and Distal Myopathy	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	NGS NGS with CNV	1300 1700
	2	MTM1, MTMR1	MLPA	590

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Cornelia de Lange Syndrome	9	AFF4, ANKRD11, HDAC8, KMT2A, NIPBL, RAD21, SMC3, SMC1A, TAF6	NGS NGS with CNV	1030 1400
	1	NIPBL	MLPA	590
Craniosynostosis	7	AFF4, ANKRD11, HDAC8, KMT2A, NIPBL, RAD21, SMC3, SMC1A, TAF6	NGS NGS with CNV	1030 1400
	1	TWIST1	MLPA	310
Dystonia	40	ACTB, ADCY5, ANO3, ARSA, ATM, ATP1A3, ATP7B, CACNA1B, CIZ1, COL6A3, DRD2, GCDH, GCH1, GNAL, GNAO1, HPCA, KCNMA1, KCTD17, MECR, PANK2, PLA2G6, PNKD, PRKN, PRKRA, PRRT2, RELN, SGCE, SLC2A1, SLC6A3, SLC25A1, SLC30A10, SLC39A14, SPR, TAF1, TBCE, TH, THAP1, TIMM8A, TOR1A, TUBB4A	NGS NGS with CNV	1300 1700
	7	ATP1A3, GCH1, PRKRA, SGCE, TH, THAP1, TOR1A	MLPA	710
Epilepsy	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	NGS NGS with CNV	1300 1700
	11	CHRNA4, CHRNB2, EPM2A, KCNQ1, KCNQ3, NHLRC1, PCDH19, SCN1A, SLC2A1, STXBP1, WWOX	MLPA	1300

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Fragile X Syndrome	1	FMR1	Repeat Expansion/ Fragment Length Analysis	262
Frontotemporal Dementia	22	ABCA7, APOE, APP, CHMP2B, CSF1R, FUS, GRN, ITM2B, MAPT, PRNP, PSEN1, PSEN2, SIGMAR1, SNCA, SORL1, TARDBP, TBK1, TREM2, TUBA4A, UBE3A, UBQLN2, VCP	NGS NGS with CNV	1030 1400
	3	CRHR1, GRN, MAPT	MLPA	590
Hereditary Spastic Paraplegia	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	NGS NGS with CNV	1030 1400
	1/1	MT-ATP6	Sanger Sequencing	87
	5	ATL1, NIPA1, SPAST, SPG7, REEP1	MLPA	806
Joubert Syndrome	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	NGS NGS with CNV	1030 1400
Leukodystrophy and Leukoencephalopathy	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	NGS NGS with CNV	1030 1400
	7	ABCD1, ASPA, L2HGDH, LMNB1, MLC1, NOTCH3, PLP1	MLPA	1070
Limb-Girdle Muscular Dystrophy	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	NGS NGS with CNV	1030 1400
	12	ANO5, CAPN3, DYSF, FKRP, LCAV3, MNA, MYOT, SGCA, SGCB, SGCD, SGCG, ZMPSTE24	MLPA	1300
Menkes Disease	1	ATP7A	Sanger Sequencing	1030
	1	ATP7A	MLPA	310
Microcephaly	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	NGS NGS with CNV	1300 1400

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Mitochondrial Diseases	37	Mitochondrial Genome	NGS	803
	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	NGS NGS with CNV	1030 1400
	10	DGUOK, MPV17, POLG, POLG2, RRM2B, SLC25A4, SUCLA2, SUCLG1, TK2, TWNK	MLPA	710
Mitochondrial Encephalomyopathy Lactic Acidosis and Stroke-Like Episodes (MELAS Syndrome)	1/1	MT-TL1	Sanger Sequencing	87
Neurodegeneration with Brain Iron Accumulation	10	ATP13A2, COASY, C19orf12, CP, DCAF17, FA2H, FTL, PANK2, PLA2G6, WDR45	NGS NGS with CNV	1030 1400
	2	PANK2, PLA2G6	MLPA	590
Parkinson's Disease	40	ADH1C, ATP1A3, ATP13A2, ATP6AP2, ATXN2, CHCHD2, DCTN1, DNAJC6, DNAJC13, EIF4G1, FBXO7, FTL, GBA, GCH1, GIGYF2, HTRA2, LRRK2, MAPT, PARK7, PINK1, PLA2G6, PODXL, PRKN, PRKRA, PTRHD1, RAB39B, SLC6A3, SLC30A10, SNCA, SNCB, SPG11, SPR, SYNJ1, TAF1, TBP (excluding exon 3), TH, TMEM230, UCHL1, VPS35, VPS13C	NGS NGS with CNV	1300 1700
	7	GCH1, LRRK2, PARK7, PINK1, PRKN, SNCA, UCHL1	MLPA	710
Short Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency	1	ACADS	Sanger Sequencing	515

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Smith-Lemli-Opitz Syndrome	1	DHCR7	Sanger Sequencing	525
Spinocerebellar Ataxias	81	ABCB7, ABHD12, ACO2, AFG3L2, ANO10, APTX, ATCAY, ATG5, ATM, ATP8A2, ATP2B3, CACNA1G, CASK, CCDC88C, CLCN2, CLN5, COQ8A, CWF19L1, CYP27A1, DARS2, FXN, GOSR2, GRID2, GRM1, DNMT1, EEF2, ELOVL4, ELOVL5, FAT2, FGF14, FLVCR1, ITPR1, KCNC3, KCND3, KCNJ10, LAMA1, MECR, MME, NOP56, NPC1, NPC2, OPHN1, PDYN, PHYH, PNKP, PNPLA6, POLG, PRKCG, RUBCN, PLD3, PTF1A, PUM1, RNF216, SACS, SCYL1, SETX, SIL1, SLC9A1, SLC9A6, SLC52A2, SNX14, SPTBN2, STUB1, SYNE1, SYT14, TRPC3, TTBK2, TDP1, TGM6, TMEM240, TPP1, TTPA, TUBB4A, TWNK, UBA5, VPS13D, VWA3B, WFS1, WWOX, XRCC1, ZNF592	NGS NGS with CNV	1300 1700
	13	ATXN1, ATXN2, ATXN3, ATXN7, ATXN8, ATXN10, ATN1, BEAN1, CACNA1A, FXN, NOP56, PPP2R2B, TBP	Repeat Expansion Analysis	3480
Tuberous Sclerosis	2	TSC1, TSC2	NGS	803
	2	TSC1, TSC2	MLPA	590
Very Long Chain Acyl-Coenzyme A Dehydrogenase (VLCAD) Deficiency	1	ACADVL	NGS	803
	1	ACADVL	MLPA	310
Wilson Disease	1	ATP7B	Sanger Sequencing	773
	1	ATP7B	MLPA	310
Whole Exome Sequencing (WES)		Solo sample	NGS NGS with CNV	1300 1700
		Trio samples	NGS NGS with CNV	2600 3400
Whole Genome Sequencing (WGS)		Solo sample Trio samples	NGS	3635 7449

Version: 2019-09-20

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 10 weeks and WGS 15 weeks

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

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