ASPER OPHTHALMICS

# Asper Ophthalmics

| Disease/Condition name | Method | No of detectable genes/markers | Genes | Price (EUR) |
|---|---|---|---|---|
| Achromatopsia | NGS | 6 | ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H | 1051 |
| Age-related Macular Degeneration | NGS | 21 | ABCA4, ARMS2, C2, C3, C9, CCR3, CFB, CFH, CFI, CST3, CXCL8, CX3CR1, ERCC6, FBLN5, HMCN1, HTRA1, IL6, IL1A, NLRP3, RAX2, TLR4 | 995* |
| | Sanger Sequencing | 2/3 | ARMS2, CFH | 87 |
| Aniridia | Sanger Sequencing | 1 | PAX6 | 773 |
| Anophthalmia/ Microphthalmia/ Coloboma/Anterior Segment Dysgenesis | NGS | 35 | ABCB6, ALDH1A3, ASPH, BCOR, B3GLCT, BMP4, CHD7, COL4A1, CYP1B1, EYA1, FOXC1, FOXE3, GDF3, GDF6, HCCS, HESX1, HMGB3, LTBP2, MAB21L2, MFRP, OTX2, PAX6, PITX2, PRSS56, RARB, RAX, SIX6, SLC38A8, SMOC1, SOX2, STRA6, TENM3, VAX1, VSX1, VSX2 | 1314 |
| Autosomal Dominant Optic Atrophy | Targeted regions sequencing by NGS | 1 | OPA1 | 450 |
| | NGS | 3 | OPA1, OPA3, TMEM126A (autosomal recessive optic atrophy) | 1030 |
| Autosomal Dominant Retinitis Pigmentosa | Targeted regions sequencing by NGS | 16 | CA4, CRX, FSCN2, IMPDH1, KLHL7, NR2E3, NRL, PRPF3, PRPF8, PRPF31, PRPH2, RHO, ROM1, RP1, RP9, TOPORS | 450 |
| | NGS | 26 | AIPL1, BEST1, CA4, CRX, FSCN2, GUCA1B, IMPDH1, KLHL7, NR2E3, NRL, PRKCG, PRPF3, PRPF6, PRPF8, PRPF31, PRPH2, RDH12, RGR, RHO, ROM1, RP1, RP9, RPE65, SEMA4A, SNRNP200, TOPORS | 1314 |
| Autosomal Recessive Retinitis Pigmentosa | Targeted regions sequencing by NGS | 28 | ABCA4, AIPL1, CERKL, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CRB1, EYS, GRK1, IMPG2, LRAT, MERTK, NR2E3, PDE6A, PDE6B, PROM1, RBP3, RDH12, RGR, RHO, RLBP1, RP1, RPE65, SAG, TULP1, USH2A | 450 |
| | NGS | 58 | ABCA4, AIPL1, ARL6, BEST1, C2orf71, C8ORF37, CA4, CERKL, CLRN1, CNGA1, CNGB1, CRB1, CRX, DHDDS, EYS, FAM161A, FLVCR1, FSCN2, GUCA1B, IDH3B, IMPDH1, IMPG2, KLHL7, LRAT, MAK, MERTK, NR2E3, NRL, PDE6A, PDE6B, PDE6G, PRCD, PROM1, PRPF3, PRPF6, PRPF8, PRPF31, PRPH2, RBP3, RDH12, RGR, RHO, RLBP1, ROM1, RP1, RP2, RP9, RPE65, RPGR (ORF15 excluded), SAG, SEMA4A, SPATA7, TOPORS, TTC8, TULP1, USH2A, ZNF513 | 1314 |
| | Sanger Sequencing | 1 | RPE65 | 515 |

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ASPER OPHTHALMICS

# Asper Ophthalmics

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|---|---|---|---|---|
| Bardet-Biedl Syndrome, McKusick-Kaufman Syndrome, Borjeson-Forssman-Lehmann Syndrome, Alstrom Syndrome, Albright Hereditary Osteodystrophy | Targeted regions sequencing by NGS | 16 | ALMS1 (excluding exon 8), ARL6, BBS1, BBS2, BBS4, BBS5, BBS7, BBS9, BBS10, BBS12, GNAS, MKKS, MKS1, PHF6, TRIM32, TTC8 | 450 |
| | NGS | 22 | ALMS1, ARL6, BBS1, BBS2, BBS4, BBS5, BBS7, BBS9, BBS10, BBS12, CCDC28B, CEP290, GNAS, LZTFL1, MKS1, MKKS, PHF6, SDCCAG8, TMEM67, TRIM32, TTC8, WDPCP | 1314 |
| Cataract | NGS | 44 | AGK, BCOR, BFSP1, BFSP2, CHMP4B, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, CTDP1, EPHA2, EYA1, FTL, FYCO1, GALK1, GCNT2, GJA1, GJA3, GJA8, HSF4, LIM2, LSS, MAF, MIP, NHS, P3H2, PAX6, PITX3, PXDN, SIPA1L3, SIL1, SIX6, SLC16A12, TDRD7, UNC45B, VIM, VSX2 | 1314 |
| Choroideremia | Sanger Sequencing | 1 | CHM | 773 |
| Cone-Rod Dystrophy | NGS | 32 | ABCA4, ADAM9, AIPL1, BEST1, CABP4, CACNA1F, CACNA2D4, CDHR1, CERKL, CNGB3, CNNM4, C8ORF37, CRX, GNAT2, GUCA1A, GUCY2D, KCNV2, PDE6C, PDE6H, PITPNM3, PROM1, PRPH2, RAB28, RAX2, RDH5, RGS9, RGS9BP, RIMS1, RPGR (ORF15 excluded), RPGRIP1, SEMA4A, UNC119 | 1314 |
| Congenital Stationary Night Blindness | NGS | 13 | CABP4, CACNA1F, CHM, GNAT1, GRK1, GRM6, NYX, PDE6B, RDH5, RHO, SAG, SLC24A1, TRPM1 | 1051 |
| Congenital Stationary Night Blindness | NGS | 13 | CABP4, CACNA1F, CHM, GNAT1, GRK1, GRM6, NYX, PDE6B, RDH5, RHO, SAG, SLC24A1, TRPM1 | 1051 |
| Corneal Dystrophy | NGS | 21 | CHST6, COL5A1, COL17A1, COL8A2, CYP4V2, DCN, GSN, KRT3, KRT12, LOXHD1, PIKFYVE, PRDM5, SLC4A11, SOD1, ZEB1, ZNF469, TACSTD2, TCF4, TGFBI, UBIAD1, VSX1 | 1051 |
| Leber Congenital Amaurosis | Targeted regions sequencing by NGS | 15 | AIPL1, CEP290, CRB1, CRX, GUCY2D, IQCB1, LCA5, LRAT, MERTK, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1 | 450 |
| | NGS | 20 | AIPL1, CABP4, CEP290 (intronic position c.2991+1655A>G included), CRB1, CRX, GDF6, GUCY2D, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, NMNAT1, OTX2, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1 | 1051 |
| Leber Hereditary Optic Neuropathy | RFLP | 3/3 | MT-ND1, MT-ND4, MT-ND6 | 176 |
| Norrie Disease | Sanger Sequencing | 1 | NDP | 262 |

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ASPER OPHTHALMICS

# Asper Ophthalmics

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|---|---|---|---|---|
| Oculocutaneous Albinism, Ocular Albinism, Hermansky-Pudlak Syndrome, Chediak-Higashi Syndrome | NGS | 17 | AP3B1, BLOC1S3, BLOC1S6, C10orf11, DTNBP1, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, OCA2, SLC24A5, SLC45A2, TYR, TYRP1 | 1051 |
| Papillorenal Syndrome | Sanger Sequencing | 1 | PAX2 | 525 |
| Retinoblastoma | NGS | 1 | RB1 | 960 |
| Stargardt Disease | NGS | 1 | ABCA4 | 450 |
| | NGS | 4 | ABCA4, CNGB3, ELOVL4, PROM1 | 1030 |
| Usher Syndrome | Targeted regions sequencing by NGS | 9 | CDH23, CLRN1, DFNB31, GPR98, MYO7A, PCDH15, USH2A, USH1C, USH1G | 450 |
| | NGS | 20* | ABHD12, CDH23, CIB2, CLRN1, COL4A6, DFNB31, DSPP (excluding exon 5), GIPC3, GPR98, HARS, KARS, LHFPL5, LOXHD1, MYO7A, PCDH15, PDZD7, TNC, USH2A, USH1C, USH1G | 1051 |
| Vitelliform Macular Dystrophy | Sanger Sequencing | 1 | BEST1 | 395 |
| | NGS | 2 | BEST1, PRPH2 | 1030 |
| X-Linked Retinitis Pigmentosa | Targeted regions sequencing by NGS, Sanger Sequencing | 2 | RP2, RPGR (ORF15 included) | 640 |
| | Sanger Sequencing | 1 | RPGR (ORF15 region only) | 257 |
| | NGS, Sanger Sequencing | 3 | OFD1, RP2, RPGR (ORF15 included) | 1051 |
| X-Linked Retinoschisis | Sanger Sequencing | 1 | RS1 | 395 |
| Eye Diseases | NGS | 277 | 277 genes associated with different eye diseases | 1567 |
| Whole Exome Sequencing (WES) | NGS | | Solo sample<br>Trio samples | 1567<br>3100 |
| Whole Genome Sequencing (WGS) | NGS | | Solo sample<br>Trio samples | 4500<br>8800 |

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* Clinical interpretation is not available

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ASPER
REPROGENETICS

# Asper Reprogenetics

| Disease/Condition name | Method | No of detectable genes/markers | Genes | Price (EUR) |
|---|---|---|---|---|
| Ashkenazi Jewish Diseases | Targeted mutation analysis by APEX | 31/117 | ABCC8, AGL, ASPA, BBS2, BCKDHB, BLM, BRCA1, BRCA2, CFTR, CLRN1, DHDDS, DLD, FAM161A, F11, FANCC, GBA, GJB2, G6PC, HEXA, IKBKAP, LCA5, LDLR, MAK, MCOLN1, MEFV, NEB, PCDH15, SERPINA1, SMPD1, TMEM216, TOR1A | 316 |
| | NGS | 33 | ABCC8, AGL, ASPA, BCKHDB, BLM, BRCA1, BRCA2, CFTR, CLRN1, CYP21A2, DLD, F11, FANCC, FKTN, GBA, GJB2, G6PC, HEXA, IKBKAP, LCA5, LDLR, LRRK2, MCOLN1, MEFV, MSH2, MSH6, NEB, PCDH15, SERPINA1, SMN1, SMPD1, TMEM216, TOR1A | 1051 |
| Carriership | Targeted mutation analysis by APEX | 20/96 | ACADM, ATP7B, CFTR, CHRNE, DHCR7, F2, F5, FAH, FSHR, HBB, HEXA, HFE, GALT, GJB2, MEFV, MTHFR, PAH, SERPINA1, SLC26A4, TCN2 | 359 |
| | NGS TruSight™ Inherited Disease | 550 | 550 genes associated with inherited diseases | 952 |
| Cystic Fibrosis | Targeted mutation analysis by APEX | 1/289 | CFTR | 316 |
| | Targeted regions sequencing by NGS | 1 | CFTR | 450 |
| Fragile X Syndrome | Repeat Expansion/ Fragment Length Analysis | 1 | FMR1 | 262 |
| Folate-Dependent Neural Tube Defects | Targeted mutation analysis | 1 | MTHFR | 93 |
| Hutterite Genetic Diseases | Targeted mutation analysis by APEX | 28/31 | ABCC8, ABCG8, ALPL, BBS2, BCHE, CFTR, CPT1A, DNAJC19, DPH1, EMG1, FKRP, GJB2, MUT, MYO7A, NDUFS4, NPHP1, PCDH15, PROP1, SLC5A5, SLC39A8, TECR, TH, TMEM237, TRIM32, TYR, VLDLR, ZMPSTE24 | 235* |
| Male Factor Infertility | Targeted mutation analysis by APEX, PCR | 19/291 | CFTR, DDX25, DNAH5, DNAH11, DNAI1, ESR2, FSHB, GNRHR, INSL3, NLRP14, PRDM9, PRM1, PRM2, PRM3, RBMXL2, RXFP2, TEKT2, USP26, UTP14C<br>47XXY<br>AZF deletions | 492 |
| | PCR | N/A | 47XXY | 87 |
| | PCR | 3/8 (+9 for extended analysis) | AZF deletions | 87 |

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ASPER
REPROGENETICS

# Asper Reprogenetics

| Disease/Condition name | Method | No of detectable genes/markers | Genes | Price (EUR) |
|---|---|---|---|---|
| Maternal Cell Contamination | Fragment Analysis | NA/6 | | 87 |
| Whole Exome Sequencing (WES) | NGS | | Solo sample<br>Trio samples | 1567<br>3100 |
| Whole Genome Sequencing (WGS) | NGS | | Solo sample<br>Trio samples | 4500<br>8800 |

* Clinical interpretation is not available

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ASPER ONCOGENETICS

# Asper Oncogenetics

| Disease/Condition name | Method | No of detectable genes/markers | Genes | Price (EUR) |
|---|---|---|---|---|
| Breast and Ovarian/ Endometrial Cancer | Targeted mutation analysis by APEX | 5/87 | BRCA1, BRCA2, CHEK2, RAD51, NBN | 316 |
| | NGS | 33 | ATM, BARD1, BRCA1, BRCA2, BRIP1, CASP8, CDH1, CHEK2, FANCA, FANCC, FANCD2 (excluding exon 15), FANCE, FANCF, FANCG, KRAS, MAP3K1, MEN1, MLH1, MSH2, MSH6, MRE11A, MUTYH, NBN, PALB2, PTCH1, PTEN, RAD50, RAD51C, RAD51D, STK11, TGFB1, TP53, XRCC2 | 1051 |
| Cancer Predisposition | NGS | 92 | AIP, ALK, APC, ATM, BAP1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BUB1B, CDC73, CDH1, CDK4, CDKN1C, CDKN2A, CEBPA, CEP57, CHEK2, CYLD, DDB2, DICER1, DIS3L2, EGFR, EPCAM, ERCC2, ERCC3, ERCC4, ERCC5, EXT1, EXT2, EZH2, FANCA, FANCB, FANCC, FANCD2 (excluding exon 15, 16), FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FH, FLCN, GATA2, GPC3, HNF1A, HRAS, KIT, MAX, MEN1, MET, MLH1, MSH2, MSH6, MUTYH, NBN, NF1, NF2, NSD1, PALB2, PHOX2B, PRF1, PRKAR1A, PTCH1, PTEN, RAD51C, RAD51D, RB1, RECQL4, RET, RHBDF2, RUNX1, SBDS, SDHAF2, SDHB, SDHC, SDHD, SLX4, SMAD4, SMARCB1, STK11, SUFU, TMEM127, TP53, TSC1, TSC2, VHL, WRN, WT1, XPA, XPC | 1051 |
| Familial Adenomatous Polyposis | Sanger Sequencing | 1 | APC | 773 |
| Fanconi Anemia | NGS | 17 | BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, PALB2, RAD51C, SLX4, XRCC2 | 1051 |
| Lynch Syndrome | Sanger Sequencing | 1<br>1<br>1 | MLH1<br>MSH2<br>MSH6 | 515<br>515<br>515 |
| | NGS | 3 | MLH1, MSH2, MSH6 | 1030 |
| | MLPA | 2/NA | MLH1, MSH2 | 257 |
| Microsatellite instability | Fragment Analysis | NA/6 | | 175 |
| MUTYH-associated Polyposis | Sanger Sequencing | 1 | MUTYH | 257 |
| | RFLP | 1/2 | MUTYH | 170 |
| Nijmegen Breakage Syndrome | Sanger Sequencing | 1<br>1/1 | NBN<br>NBN | 788<br>90 |

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ASPER
ONCOGENETICS

# Asper Oncogenetics

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|---|---|---|---|---|
| Polyposis Syndromes | NGS | 6 | APC, BMPR1A, MUTYH, PTEN, SMAD4, STK11 | 1030 |
| Thyroid Cancer | NGS | 10 | APC, CDC73, DICER1, MEN1, PRKAR1A, PTEN, SDHB, SDHD, RET, TP53 | 1051 |
| Whole Exome Sequencing (WES) | NGS | | Solo sample<br>Trio samples | 1567<br>3100 |
| Whole Genome Sequencing (WGS) | NGS | | Solo sample<br>Trio samples | 4500<br>8800 |

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ASPER
CARDIOGENETICS

# Asper Cardiogenetics

| Disease/Condition name | Method | No of detectable genes/markers | Genes | Price (EUR) |
|---|---|---|---|---|
| Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy | NGS | 14 | CTNNA3, DES, DSG2, DSC2, DSP, JUP, LDB3, LMNA, PKP2, PLN, RYR2, TGFB3, TMEM43, TTN | 1070 |
| Brugada Syndrome | NGS | 15 | CACNA1C, CACNA2D1, CACNB2, GPD1L, HCN4, KCND3, KCNE3, KCNE1L, KCNJ8, RANGRF, SCN5A, SCN1B, SCN2B, SCN3B, TRPM4 | 1314 |
| Familial Hypercholesterolemia | NGS | 4 | APOB, LDLR, LDLRAP1, PCSK9 | 1030 |
| Familial Thoracic Aortic Aneurysm and Dissection and Related Syndromes | NGS | 12 | ACTA2, COL3A1, COL5A1, FBN1, MYH11, MYLK, SLC2A10, SMAD3, TGFB2, TGFBR1, TGFBR2, TGFBR3 | 1288 |
| Hyperlipidemia | Sanger Sequencing | 1/2 | APOE | 87 |
| Hypertrophic Cardiomyopathy | NGS | 28 | ACTC1, ACTN2, CALR3, CAV3, CSRP3, GLA, JPH2, LAMP2, LDB3, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, MYPN, NEXN, PLN, PRKAG2, SLC25A4, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTR, VCL | 1314 |
| Long QT Syndrome | NGS | 14 | AKAP9, ANK2, CACNA1C, CALM1, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN5A, SCN4B, SNTA1 | 1051 |
| Noonan Syndrome | NGS | 13 | BRAF, CBL, HRAS, KAT6B, KRAS, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, SHOC2, SOS1, SPRED11 | 1051 |
| Pulmonary Arterial Hypertension | NGS, Sanger Sequencing | 7 | ACVRL1, BMPR2, BMPR1B, CAV1, ENG, KCNK3, SMAD9 | 1051 |
| Statin-Induced Myopathy | Sanger Sequencing | 1/1 | SLCO1B1 | 87 |
| Venous Thrombosis | RFLP | 3/4 | F2, F5, MTHFR | 87 |
| Whole Exome Sequencing (WES) | NGS | | Solo sample<br>Trio samples | 1567<br>3100 |
| Whole Genome Sequencing (WGS) | NGS | | Solo sample<br>Trio samples | 4500<br>8800 |

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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 |
| 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 | 1/NA | PMP22 | 316 |
| Cornelia de Lange Syndrome | NGS | 5 | HDAC8, NIPBL, RAD21, SMC3, SMC1A | 1030 |
| Craniosynostosis | NGS, Sanger Sequencing | 7 | FGFR1, FGFR2, FGFR3, IL11RA, MSX2, RECQL4, TWIST1 | 1051 |
| 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 |
| Epilepsy | NGS | 127 | AARS, ADAR, ADSL, ALDH7A1, ALG3, ALG13, ARHGEF9, ARX, ATP1A2, ATP1A3, ATP6AP2, ATRX, BRAT1, CACNA1A, CACNA1D, CACNA1H, CACNB4, CASK, CDKL5, CERS1, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN2, CPA6, CSTB, DEPDC5, DNM1, DOCK7, EEF1A2, EFHC1, EPM2A, FGF12, FLNA, FOXG1, GABRA1, GABRB3, GABRD, GABRG2, GAMT, GATM, GNAO1, GOSR2, GPHN, GRIN1, GRIN2A, GRIN2B, HCN1, HUWE1, ITPA, IQSEC2, KCNA1, KCNA2, KCNB1, KCNC1, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCTD7, KIAA2022, KIF1A, KIF5C, LGI1, MBD5, MCCC1, MECP2, MEF2C, MOCS1, MOCS2, MTOR, NECAP1, NHLRC1, NRXN1, PCDH19, PIK3R2, PIGA, PIGO, PIGT, PLCB1, PNKP, PNPO, POLG, PRICKLE1, PRRT2, PURA, RELN, ROGDI, SCARB2, SCN1A, SCN1B, SCN2A, SCN8A, SERPINI1, SIK1, SLC12A5, SLC13A5, SLC25A22, SLC2A1, SLC35A2, SLC35A3, SLC6A1, SLC6A8, SLC9A6, SMARCA2, SNIP1, SPATA5, SPTAN1, SRPX2, ST3GAL3, ST3GAL5, STX1B, STXBP1, SYN1, SYNGAP1, SYP, SZT2, TBC1D24, TCF4, TSC1, TSC2, TUBB3, UBE3A, WDR45, WWOX, ZDHHC9 | 1546 |

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ASPER
NEUROGENETICS

# Asper Neurogenetics

| Disease/Condition name | Method | No of detectable genes/markers | Genes | Price (EUR) |
|---|---|---|---|---|
| Fragile X Syndrome | Repeat Expansion/ Fragment Length Analysis | 1 | FMR1 | 262 |
| Frontotemporal Dementia | NGS | 5 | CHMP2B, GRN, MAPT, TARDBP, PSEN1 | 1051 |
| Hereditary Spastic Paraplegia | NGS | 34 | ATL1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, B4GALNT1, BSCL2, CYP7B1, CYP2U1, DDHD2, ERLIN2, FA2H, GBA2, GJC2, HSPD1, KIAA0196, KIF1A, KIF5A, L1CAM, NIPA1, PLP1, PNPLA6, REEP1, RTN2, SLC16A2, SPAST, SPG7, SPG11, SPG20, SPG21, TECPR2, VPS37A, ZFYVE26 | 1314 |
| | Sanger Sequencing | 1/1 | MT-ATP6 | 87 |
| 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 |
| Menkes Disease | Sanger Sequencing | 1 | ATP7A | 1030 |
| 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 | Sanger Sequencing/ NGS | 37 | Mitochondrial Genome | 1030 |
| | NGS | 133 | AARS2, ABCB7, ACAD9, ACADL, ACADM, ACADS, ACADVL, ADCK3, AFG3L2, AIFM1, ALAS2, APTX, ATP5E, ATPAF2, AUH, BCS1L, BOLA3, C10orf2, C12orf65, CISD2, COA5, COQ2, COQ6, COQ9, 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 | 1314 |

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| Mitochondrial Diseases | NGS | 133 | SLC37A4, SOD1, SPG7, SUCLA2, SUCLG1, SURF1,TACO1, TAZ, TIMM8A, TK2, TMEM126A, TMEM70, TRMU, TSFM, TTC19, TUFM, TYMP, UQCRB, UQCRQ, WFS1, YARS2 | 1314 |
| 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 |
| Parkinson Disease | NGS | 20 | ADH1C, ATP13A2, ATXN2, DNAJC6, EIF4G1, FBXO7, GBA, GIGYF2, HTRA2, LRRK2, MAPT, PARK2, PARK7, PINK1, PLA2G6, SLC6A3, SNCA, TBP (excluding exon 3), UCHL1, VPS35 | 1314 |
| Short Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency | Sanger Sequencing | 1 | ACADS | 515 |
| Smith-Lemli-Opitz Syndrome | Sanger Sequencing | 1 | DHCR7 | 525 |
| Spinocerebellar Ataxias | NGS | 64 | ABCB7, ABHD12, ACO2, ADCK3, AFG3L2, ANO10, APTX, ATCAY, ATM, ATP8A2, CASK, CCDC88C, CLCN2, CLN5, C10orf2, CWF19L1, CYP27A1, DARS2, DNMT1, EEF2, ELOVL4, ELOVL5, FGF14, FLVCR1, FXN, GOSR2, GRM1, ITPR1, KCNC3, KCND3, KCNJ10, LAMA1, NOP56, OPHN1, PDYN, PHYH, PNKP, PNPLA6, POLG, PRKCG, PTF1A, RNF216, RUBCN, SACS, SETX, SIL1, SLC9A1, SLC9A6, SLC52A2, SNX14, SPTBN2, STUB1, SYNE1, SYT14, TTBK2, TDP1, TGM6, TMEM240, TPP1, TTPA, TUBB4A, WFS1, WWOX, ZNF592 | 1314 |
| | Repeat Expansion Analysis | 12 | ATXN1, ATXN2, ATXN3, ATXN7, ATXN8OS, ATXN10, ATN1, BEAN1, CACNA1A, FXN, NOP56, PPP2R2B, TBP | 2677 |
| Very Long Chain Acyl-Coenzyme A Dehydrogenase (VLCAD) Deficiency | Sanger Sequencing | 1 | ACADVL | 515 |
| Wilson Disease | Targeted mutation analysis by APEX | 1/117 | ATP7B | 316 |
| | Sanger Sequencing | 1 | ATP7B | 773 |

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|---|---|---|---|---|
| Whole Exome Sequencing (WES) | NGS | | Solo sample<br>Trio samples | 1567<br>3100 |
| Whole Genome Sequencing (WGS) | NGS | | Solo sample<br>Trio samples | 4500<br>8800 |

**Service includes**
- DNA extraction
- Genotyping
- Bioinformatic analysis of detected variants
- Detected variants available in Excel format (on request)
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ASPER
OTOGENETICS

# Asper Otogenetics

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|---|---|---|---|---|
| Alport Syndrome | NGS | 3 | COL4A3, COL4A4, COL4A5 | 1030 |
| Aminoglycoside-Induced Deafness | Sanger sequencing | 1/1 | MT-RNR1 | 87 |
| Branchiootorenal Syndrome | NGS | 3 | EYA1, SIX1, SIX5 | 1288 |
| Jervell and Lange-Nielson Syndrome | Sanger Sequencing | 2 | KCNE1, KCNQ1 | 773 |
| Palmoplantar Keratoderma with Deafness | Sanger Sequencing | 1 | GJB2 | 257 |
| Pendred Syndrome | Sanger Sequencing | 1 | SLC26A4 | 773 |
| Sensorineural Hearing Loss | Targeted regions sequencing by NGS | 11 | GJB2, GJB3, GJB6, KCNQ4, MYO7A, MYO15A, MT-RNR1, MT-TS1, SLC26A4, SLC26A5, TMC1 | 450 |
| | NGS | 76 | ACTG1, ATP2B2, ATP6V1B1, BSND, CCDC50, CDH23, CEACAM16, CIB2, CLDN14, CLRN1, COCH, COL11A2, CRYM, DFNA5, DFNB31, DFNB59, DIABLO, DIAPH1, DIAPH3, ESPN, ESRRB, EYA4, FOXI1, GIPC3, GJB2, GJB3, GJB6, GPR98, GPSM2, GRHL2, GRXCR1, HGF, ILDR1, KCNJ10, KCNQ4, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MIR96, MSRB3, MYH14, MYH9, MYO15A, MYO1A, MYO3A, MYO6, MYO7A, OTOA, OTOF, PCDH15, PDZD7, POU3F4, POU4F3, PRPS1, PTPRQ, RDX, SERPINB6, SIX1, SLC17A8, SLC26A4, SLC26A5, SMPX, STRC, TECTA, TJP2, TMC1, TMIE, TMPRSS3, TPRN, TRIOBP (excluding exon 7), TRMU, USH2A, USH1C, USH1G, WFS1 | 1314 |
| | Sanger Sequencing | 1 | GJB2 | 257 |
| Stickler Syndrome | NGS | 6 | COL2A1, COL11A1, COL11A2, COL9A1, COL9A2, COL9A3 | 1030 |
| Treacher-Collins Syndrome | NGS | 3 | POLR1C, POLR1D, TCOF1 | 1030 |
| Usher Syndrome | Targeted regions sequencing by NGS | 9 | CDH23, CLRN1, DFNB31, GPR98, MYO7A, PCDH15, USH2A, USH1C, USH1G | 450 |
| | NGS | 20 | ABHD12, CDH23, CIB2, CLRN1, COL4A6, DFNB31, DSPP, GIPC3, GPR98, HARS, KARS, LHFPL5, LOXHD1, MYO7A, PCDH15, PDZD7, TNC, USH2A, USH1C, USH1G | 1051 |
| Waardenburg Syndrome | NGS | 6 | EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10 | 1030 |

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|---|---|---|---|---|
| Zellweger Spectrum Disorders | NGS | 14 | PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PHYH | 1051 |
| Whole Exome Sequencing (WES) | NGS | | Solo sample<br>Trio samples | 1567<br>3100 |
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ASPER
HEMATOLOGY

# Asper Hematology

| Disease/Condition name | Method | No of detectable genes/markers | Genes | Price (EUR) |
|---|---|---|---|---|
| Alpha-Thalassemia | PCR | 2/7 | HBA1, HBA2 | 91 |
| Beta-Thalassemia, Sickle Cell Disease | Sanger Sequencing | 1 | HBB | 257 |
| Fanconi Anemia | NGS | 17 | BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, PALB2, RAD51C, SLX4, XRCC2 | 1051 |
| Thrombocytopenia | NGS | 14 | ADAMTS13, ANKRD26, CYCS, GATA1, GP1BA, GP1BB, GP9, ITGA2B, ITGB3, MASTL, MPL, MYH9, RUNX1, WAS | 1070 |
| Whole Exome Sequencing (WES) | NGS | | Solo sample<br>Trio samples | 1567<br>3100 |
| Whole Genome Sequencing (WGS) | NGS | | Solo sample<br>Trio samples | 4500<br>8800 |

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ASPER
DYSMORPHOLOGY

# Asper Dysmorphology

| Disease/Condition name | Method | No of detectable genes/markers | Genes | Price (EUR) |
|---|---|---|---|---|
| Brain malformations | NGS | 147 | ACTB, ACTG1, ADGRG1, AHI1, AKT3, AMPD2, AMT, AP4M1, ARFGEF2, ARL13B, ARX, ASPM, ATP6V0A2, ATR, ATRX, B9D1, B3GALNT2, B4GAT1, CASK, CC2D2A, CCND2, CDK5RAP2, CENPJ, CEP135, CEP290, CEP152, CEP63, CEP41, CHMP1A, CLP1, C5orf42, CREBBP, CUL4B, DCX, DHCR7, DHCR24, DLAT, DLD, DYNC1H1, ETFA, EFTUD2, ERMARD, ETFB, ETFDH, EXOSC3, FAT4, FKRP, FKTN, FLNA, GCSH, GLDC, GMPPB, GPSM2, IER3IP1, INPP5E, ISPD, KIF11, KIF7, KIF2A, KIF1BP, KIF5C, KNL1, LAMA2, LAMB1, LAMC3, LARGE1, MCPH1, MECP2, MKS1, NBN, NDE1, NHEJ1, NPHP1*, OCLN, OFD1, OPHN1, PAFAH1B1, PCNT, PDHA1, PDHB, PDHX, PDP1, PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PIEZO2, PIK3R2, PNKP, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PQBP1, RAB18, RAB3GAP1, RAB3GAP2, RARS2, RELN, RPGRIP1L, RTTN, SEPSECS, SLC12A6, SLC25A19, SNAP29, SRD5A3, SRPX2, STIL, TBC1D20, TCF4, TCTN1, TCTN2, TCTN3, TMEM231, TMEM237, TMEM216, TMEM138, TMEM67, TMEM5, TSEN2, TSEN34, TSEN54, TTC21B, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP6, VLDLR, VRK1, WASHC5, WDR62, ZEB2, ZNF423<br>* - 250kb deletion in the heterozygous state is not detectable with the test | 1314 |
| Craniosynostosis | NGS, Sanger Sequencing | 7 | FGFR1, FGFR2, FGFR3, IL11RA, MSX2, RECQL4, TWIST1 | 1051 |
| Jeune Syndrome | NGS | 15 | CEP120, CSPP1, DYNC2H1, DYNC2LI1, IFT52, IFT80, IFT140, IFT172, KIAA0586, NEK1, TTC21B, WDR19, WDR34, WDR35, WDR60 | 1546 |
| 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 |
| Noonan Syndrome | NGS | 13 | BRAF, CBL, HRAS, KAT6B, KRAS, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, SHOC2, SOS1, SPRED1 | 1051 |
| Skeletal Dysplasia | NGS | 15 | ALPL, COL2A1, ESCO2, FGFR1, FGFR2, FGFR3, IL11RA, MSX2, RECQL4, ROR2, SLC26A2, SOX9, TRIP11, TWIST1, WNT5A | 1051 |
| Smith-Lemli-Opitz Syndrome | Sanger Sequencing | 1 | DHCR7 | 525 |

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