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 | 15 | ABCA4, ARMS2, C2, C3, C9, CFB, CFH, CFI, CST3, CX3CR1, ERCC6, FBLN5, HMCN1, HTRA1, RAX2 | 995* |
| | Sanger Sequencing | 2/3 | ARMS2, CFH | 87 |
| Aniridia | Sanger Sequencing | 1 | PAX6 | 773 |
| | MLPA | 1 | PAX6 | 310 |
| 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 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 |
| | MLPA | 4 | IMPDH1, PRPF31, RHO, RP1 | 590 |
| 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 | 56 | ABCA4, AIPL1, ARL6, BEST1, C2orf71, C8ORF37, CA4, CERKL, CLRN1, CNGA1, CNGB1, CRB1, CRX, DHDDS, EYS, FAM161A, FLVCR1, FSCN2, GUCA1B, IDH3B, IMPDH1, IMPG2, 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 |
| | MLPA | 7 | EYS, IMPDH1, PRPF31, RHO, RP1, RP2, RPGR | 806 |
| | Sanger Sequencing | 1 | RPE65 | 515 |

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|---|---|---|---|---|
| Bardet-Biedl Syndrome, McKusick-Kaufman Syndrome, Borjeson-Forssman-Lehmann Syndrome, Alstrom Syndrome, Albright Hereditary Osteodystrophy | 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 |
| | MLPA | 1 | CHM | 310 |
| Cone-Rod Dystrophy | NGS | 35 | ABCA4, ADAM9, AIPL1, BEST1, CABP4, CACNA1F, CACNA2D4, CDHR1, CERKL, CNGB3, CNNM4, C8ORF37, CRX, GNAT2, GUCA1A, GUCY2D, KCNV2, PCYT1A, PDE6C, PDE6H, PITPNM3, POC1B, PROM1, PRPH2, RAB28, RAX2, RDH5, RGS9, RGS9BP, RIMS1, RPGR (ORF15 excluded), RPGRIP1, SEMA4A, TTLL5, UNC119 | 1314 |
| Congenital Stationary Night Blindness | NGS | 13 | CABP4, CACNA1F, CHM, GNAT1, GRK1, GRM6, NYX, PDE6B, RDH5, RHO, SAG, SLC24A1, TRPM1 | 1051 |
| Corneal Dystrophy | NGS | 20 | CHST6, COL5A1, COL17A1, COL8A2, CYP4V2, DCN, GSN, KRT3, KRT12, LOXHD1, PIKFYVE, PRDM5, SLC4A11, SOD1, ZEB1, ZNF469, TACSTD2, TGFBI, UBIAD1, VSX1 | 1051 |
| Glaucoma | NGS | 19 | ACVR1, ASB10, BEST1, CANT1, COL18A1, CYP1B1, FOXC1, LMX1B, LOXL1, LTBP2, MYOC, NTF4, OPTN, PAX6, PITX2, PITX3, SBF2, SLC4A4, WDR36 | 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 |
| | MLPA | 8 | AIPL1, CEP290, CRB1, CRX, GUCY2D, RDH12, RPE65, RPGRIP1 | 806 |

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|---|---|---|---|---|
| Leber Hereditary Optic Neuropathy | Sanger Sequencing | 3/3 | MT-ND1, MT-ND4, MT-ND6 | 176 |
| | NGS | 37 | Mitochondrial Genome | 803 |
| Norrie Disease | Sanger Sequencing | 1 | NDP | 262 |
| Oculocutaneous Albinism, Ocular Albinism, Hermansky-Pudlak Syndrome, Chediak-Higashi Syndrome | NGS | 17 | AP3B1, BLOC1S3, BLOC1S6, DTNBP1, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LRMDA, LYST, OCA2, SLC24A5, SLC45A2, TYR, TYRP1 | 1051 |
| | MLPA | 3 | GPR143, OCA2, TYR | 710 |
| Optic Atrophy | Targeted regions sequencing by NGS | 1 | OPA1 | 450 |
| | NGS | 3 | OPA1, OPA3, TMEM126A | 1030 |
| | MLPA | 1 | OPA1 | 310 |
| Papillorenal Syndrome | Sanger Sequencing | 1 | PAX2 | 525 |
| Retinoblastoma | NGS | 1 | RB1 | 960 |
| | MLPA | 1 | RB1 | 310 |
| Senior-Loken Syndrome | NGS | 9 | CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19 | 1070 |
| Stargardt Disease | NGS | 1 | ABCA4 | 450 |
| | NGS | 4 | ABCA4, CNGB3, ELOVL4, PROM1 | 1030 |
| | MLPA | 1 | ABCA4 | 590 |
| Usher Syndrome | Targeted regions sequencing by NGS | 9 | ADGRV1 (GPR98), CDH23, CLRN1, MYO7A, PCDH15, USH2A, USH1C, USH1G, WHRN (DFNB31) | 450 |
| | NGS | 20 | ABHD12, ADGRV1 (GPR98), CDH23, CIB2, CLRN1, COL4A6, DSPP (excluding exon 5), GIPC3, HARS, KARS, LHFPL5, LOXHD1, MYO7A, PCDH15, PDZD7, TNC, USH2A, USH1C, USH1G, WHRN (DFNB31) | 1051 |
| | MLPA | 1 | USH2A | 590 |
| | MLPA | 1 | PCDH15 | 310 |
| Vitelliform Macular Dystrophy | Sanger Sequencing | 1 | BEST1 | 395 |
| | NGS | 3 | BEST1, IMPG1, PRPH2 | 1030 |
| | MLPA | 2 | BEST1, PRPH2 | 590 |

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| Disease/Condition name | Method | No of detectable genes/markers | Genes | Price (EUR) |
|---|---|---|---|---|
| Vitreoretinopathy | NGS | 19 | BEST1, CAPN5, COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, COL18A1, FZD4, KCNJ13, KIF11, LRP5, NDP, NR2E3, RS1, TSPAN12, VCAN, ZNF408 | 1334 |
| 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 |
| | MLPA | 2 | RP2, RPGR | 590 |
| X-Linked Retinoschisis | Sanger Sequencing | 1 | RS1 | 395 |
| Eye Diseases | NGS | 283 | 283 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 |

* Clinical interpretation is not available

**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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