ASPER OPHTHALMICS

Asper Ophthalmics

Disease/Condition name	No of detectable genes/markers	Genes	Method	Price (EUR)
Achromatopsia	6	ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H	NGS NGS with CNV	1030 1400
Age-related Macular Degeneration	2/3	ARMS2, CFH	Sanger Sequencing	87
Aniridia	1	PAX6	Sanger Sequencing MLPA	450 310
Anophthalmia/ Microphthalmia/ Coloboma/Anterior Segment Dysgenesis	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	NGS NGS with CNV	1030 1400
Autosomal Dominant Retinitis Pigmentosa	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	NGS NGS with CNV	1030 1400
	4	IMPDH1, PRPF31, RHO, RP1	MLPA	590
	16	CA4, CRX, FSCN2, IMPDH1, KLHL7, NR2E3, NRL, PRPF3, PRPF8, PRPF31, PRPH2, RHO, ROM1, RP1, RP9, TOPORS	Targeted regions sequencing by NGS	450
Autosomal Recessive Retinitis Pigmentosa	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	NGS NGS with CNV	1030 1400
	7	EYS, IMPDH1, PRPF31, RHO, RP1, RP2, RPGR	MLPA	806
	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	Targeted regions sequencing by NGS	450
	1	RPE65	Sanger Sequencing	515

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Bardet-Biedl Syndrome, McKusick-Kaufman Syndrome, Borjeson-Forssman-Lehmann Syndrome, Alstrom Syndrome, Albright Hereditary Osteodystrophy	25	ALMS1 (excluding exon 8), ARL6, BBIP1, BBS1, BBS2, BBS4, BBS5, BBS7, BBS9, BBS10, BBS12, CCDC28B, CEP290, GNAS, IFT27, IFT172, LZTFL1, MKKS, MKS1, PHF6, SDCCAG8, TMEM67, TRIM32, TTC8, WDPCP	NGS NGS with CNV	1030 1400
Cataract	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	NGS NGS with CNV	1300 1700
Choroideremia	1	CHM	Sanger Sequencing MLPA	700 310
Cone-Rod Dystrophy	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	NGS NGS with CNV	1030 1400
Congenital Stationary Night Blindness	13	CABP4, CACNA1F, CHM, GNAT1, GRK1, GRM6, NYX, PDE6B, RDH5, RHO, SAG, SLC24A1, TRPM1	NGS NGS with CNV	1030 1400
Corneal Dystrophy	20	CHST6, COL5A1, COL17A1, COL8A2, CYP4V2, DCN, GSN, KRT3, KRT12, LOXHD1, PIKFYVE, PRDM5, SLC4A11, SOD1, ZEB1, ZNF469, TACSTD2, TGFBI, UBIAD1, VSX1	NGS NGS with CNV	1030 1400
Ectopia Lentis	18	AASS, ADAMTS10, ADAMTS17, ADAMTSL4, ASPH, BCOR, CBS, COL8A2, COL18A1, CYP1B1, FBN1, FOXC1, LTBP2, PAX6, P3H2, PORCN, SUOX, VSX2	NGS NGS with CNV	1030 1400
Glaucoma	19	ACVR1, ASB10, BEST1, CANT1, COL18A1, CYP1B1, FOXC1, LMX1B, LOXL1, LTBP2, MYOC, NTF4, OPTN, PAX6, PITX2, PITX3, SBF2, SLC4A4, WDR36	NGS NGS with CNV	1030 1400
Leber Congenital Amaurosis	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	NGS NGS with CNV	1030 1400
	8	AIPL1, CEP290, CRB1, CRX, GUCY2D, RDH12, RPE65, RPGRIP1	MLPA	806

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Leber Congenital Amaurosis	15	AIPL1, CEP290, CRB1, CRX, GUCY2D, IQCB1, LCA5, LRAT, MERTK, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1	Targeted regions sequencing by NGS	450
Leber Hereditary Optic Neuropathy	3/3	MT-ND1, MT-ND4, MT-ND6	Sanger Sequencing	176
	37	Mitochondrial Genome	NGS	803
Norrie Disease	1	NDP	Sanger Sequencing	262
Oculocutaneous Albinism, Ocular Albinism, Hermansky-Pudlak Syndrome, Chediak-Higashi Syndrome	17	AP3B1, BLOC1S3, BLOC1S6, DTNBP1, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LRMDA, LYST, OCA2, SLC24A5, SLC45A2, TYR, TYRP1	NGS NGS with CNV	1030 1400
	3	GPR143, OCA2, TYR	MLPA	710
Optic Atrophy	3	OPA1, OPA3, TMEM126A	NGS NGS with CNV	1030 1400
	1	OPA1	MLPA Targeted regions sequencing by NGS	310 450
Papillorenal Syndrome	1	PAX2	Sanger Sequencing	525
Retinoblastoma	1	RB1	NGS MLPA	960 310
Senior-Loken Syndrome	9	CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19	NGS NGS with CNV	1030 1400
Stargardt Disease	4	ABCA4, CNGB3, ELOVL4, PROM1	NGS NGS with CNV	1030 1400
	1	ABCA4	NGS MLPA	450 590
Usher Syndrome	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)	NGS NGS with CNV	1030 1400
	1	USH2A	MLPA	590
	1	PCDH15	MLPA	310
	9	ADGRV1 (GPR98), CDH23, CLRN1, MYO7A, PCDH15, USH2A, USH1C, USH1G, WHRN (DFNB31)	Targeted regions sequencing by NGS	450

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Vitelliform Macular Dystrophy	3	BEST1, IMPG1, PRPH2	NGS NGS with CNV	1030 1400
	2	BEST1, PRPH2	MLPA	590
	1	BEST1	Sanger Sequencing	395
Vitreoretinopathy	19	BEST1, CAPN5, COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, COL18A1, FZD4, KCNJ13, KIF11, LRP5, NDP, NR2E3, RS1, TSPAN12, VCAN, ZNF408	NGS NGS with CNV	1030 1400
X-Linked Retinitis Pigmentosa	3	OFD1, RP2, RPGR (ORF15 included)	NGS, Sanger Sequencing NGS with CNV	1030 1400
	2	RP2, RPGR	MLPA	590
	2	RP2, RPGR (ORF15 included)	Targeted regions sequencing by NGS, Sanger Sequencing	640
	1	RPGR (ORF15 region only)	Sanger Sequencing	257
X-Linked Retinoschisis	1	RS1	Sanger Sequencing	395
Eye Diseases	283	283 genes associated with different eye diseases	NGS NGS with CNV	1300 1700
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: 2019-06-28

* 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 10 weeks and WGS 15 weeks

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

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