

Asper Ophthalmics

Genetic test	Disease/condition name	Method	No of detectable markers	Gene	Quantity of DNA	TAT
ABCA4	Stargardt Disease (STGD), Cone-Rod Dystrophy	APEX, Sequencing	630	ABCA4	3 µg	4-6 weeks*
ADOA	Autosomal Dominant Optic Atrophy	APEX	120	OPA-1	2 µg	4-6 weeks*
ADRP	Autosomal Dominant Retinitis Pigmentosa	APEX	414	Ca4, FSCN2, IMPDH1, NRL, PRPF3, PRPF31, PRPF8, RDS, RHO, ROM1, RP1, RP9, CRX, TOPORS, PNR, KLHL7	3 µg	4-6 weeks*
AMD	Age-Related Macular Degeneration	Sequencing	6	ARMS2, CFH, CFB, C2	200 ng	4-6 weeks
ARRP	Autosomal Recessive Retinitis Pigmentosa	APEX	594	CERKL, CNGA1, CNGB1, MERTK, PDE6A, PDE6B, PNR, RDH12, RGR, RLBP1, SAG, TULP1, CRB, RPE65, USH2A, USH3A, LRAT, PROML1, PBP3	6 µg	4-6 weeks*
		Sequencing		RPE65	6 µg	4-6 weeks
BBS, MKKS, BFLS, ALMS, AHO	Bardet-Biedl Syndrome, McKusick-Kaufman Syndrome, Borjeson-Forssman-Lehmann Syndrome, Alstrom Syndrome, Albright Hereditary Osteodystrophy	APEX	346	BBS1, BBS2, BBS3, BBS4, BBS5, BBS6, BBS7, BBS8, BBS9, BBS10, BBS11, BBS12, BBS13, PHF6, ALMS1, GNAS1	5 µg	4-6 weeks*
		Sequencing		BBS1, BBS2, BBS10	5 µg	4-6 weeks
Cornea	Corneal Dystrophy	APEX	333	COL8A2, TGFB1, VSX1, CHST6, KRT3, KRT12, GSN, TACSTD2, CYP4V2, SOD1, TCF8/ZEB1, SLC4A11, UBIAD1	3 µg	4-6 weeks*
CSNB	Congenital Stationary Night Blindness	APEX	159	RHO, PDE6B, GNAT1, CABP4, GRM6, SAG, NYX, CACNA1F, CACNA2D, GRK1, TRPM1	2,5 µg	4-6 weeks*
LCA	Leber Congenital Amaurosis	APEX	641	AIPL1, CRB1, CRX, GUCY2D, LRAT, TULP1, MERTK, RPE65, RPGRIP1, CEP290, RDH12, LCA5, SPATA7	6 µg	4-6 weeks*
LHON	Leber Hereditary Optic Neuropathy	RFLP	3	MT-ND4, MT-ND6, MT-ND1	500 ng	3-4 weeks
Usher	Usher Syndrome	APEX	612	CDH23, MYO7A, PCDH15, Harmonin, SANS, USH2A, VLGR1, USH3A, Whirlin	6 µg	4-6 weeks*
VMD	Vitelliform Macular Dystrophy	APEX	138	BEST1	1 µg	4-6 weeks*
XLRP	X-Linked Retinitis Pigmentosa	APEX	184	RP2, RPGR	3 µg	4-6 weeks*

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Asper Reprogenetics

Genetic test	Disease/condition name	Method	No of detectable markers	Gene	Quantity of DNA	TAT
Ashkenazi	Ashkenazi Jewish diseases testing	APEX	78	HEXA, BLM, ASPA, SMPD1, IKBKAP, DYT1, MEFV, MCOLN1, FANCC, F11, G6PC, BCKHDB, GJB2, DLD, GDE, SERPINA1, NEB, PCDH15, ABCC8, GBA, CFTR, LDLR, TMEM216	3 µg	4-6 weeks*
Carrier	Carriership testing	RFLP	92	HBB, CFTR, HEXA, HFE, F2, F5, ACADM, GALT, GJB2, MTHFR, AAT/SERPINA1, MEFV, FAH, ATP7B, SLC26A4, TCN2, DHCR7, PAH, CHRNE, FSHr	3,5 µg	4-6 weeks*
CF	Cystic Fibrosis	APEX, Sequencing	271	CFTR	2 µg	4-6 weeks*
CAH	Congenital Adrenal Hyperplasia	APEX, MLPA	20	CYP21A2	250 ng	4-6 weeks
Noonan	Noonan Syndrome	APEX	108	PTPN11, SOS1, RAF1, KRAS, MEK1	1,5 µg	4-6 weeks
NT	Syndromes related to increased Nuchal Translucency of fetus	APEX, RFLP, MLPA, Fragment Analysis	251	CYP21A2, PTPN11, SOS1, RAF1, KRAS, MEK1, DHCR7, SMN1	4,5 µg - fetal DNA	2-3 weeks
					50 ng - maternal DNA	
SD	Skeletal Dysplasia	APEX, Sequencing	377	FGFR3, FGFR2, COL2A1, SLC26A2, ALPL, ROR2, ESCO2, CHST3, SOX9SRY	4,5 µg - fetal DNA	2-3 weeks
					50 ng - maternal DNA	
SLO	Smith-Lemli-Opitz Syndrome	APEX	121	DHCR7	420 ng	4-6 weeks
Venous Thrombosis risk assessment	Venous Thrombosis	RFLP	4	Factor V, prothrombin/ Factor II, MTHFR	300 ng	1-2 weeks

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Asper Oncogenetics

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BRAF	Colorectal Cancer, Melanoma	AS-PCR	1	BRAF	tumor tissue	2 weeks
BRCA	Hereditary Breast and Ovarian Cancer	APEX	83	BRCA1, BRCA2, CHEK2, RAD51, NBN	3 µg	4-6 weeks*
		Sequencing		BRCA1, BRCA2	3 µg	4-6 weeks
FAP	Familial Adenomatous Polyposis			APC	1,7 µg	4-6 weeks
GB	Glioblastoma	APEX	136	IDH1, IDH2, CDKN2A, EGFR, NF1, PTEN, PTPN11, RB1, TP53, TSC1, TSC2, PMS2, MLH1, MSH2, MSH6	3 µg	4-6 weeks*
HNPCC/ Lynch Syndrome	Hereditary Nonpolyposis Colorectal Cancer/Lynch Syndrome	Sequencing		MLH1, MSH2, MSH6, PMS2	2 µg	4-6 weeks
		MLPA		MLH1, MSH2	300 ng	4-6 weeks
MAP	MUTYH-Associated Polyposis	RFLP	2	MUTYH	2 µg	4-6 weeks
		Sequencing				
MMR genes methylation analysis	Colorectal Cancer	MLPA		MLH1, MSH2, MSH6, MSH3, MLH3, PMS2	tumor-, normal tissue	4-6 weeks
MMR protein expression analysis	Colorectal Cancer	IHC		MSH2, MLH1, MSH6, PMS2	tumor-, normal tissue	2 weeks
MSI	Microsatellite instability	Fragment Analysis	6		tumor-, normal tissue	3 weeks
TPMT	Thiopurine S-Methyltransferase Deficiency	RFLP	3	TPMT	300 ng	1-2 weeks

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ApoE	Recovery from traumatic brain injury, Alzheimer's Disease, coronary heart disease	Sequencing	2	ApoE	300 ng	4-6 weeks
Beta-Thalassemia	Beta-Thalassemia	APEX	69	HBB	1 µg	4-6 weeks*
Lactose Intolerance	Lactose Intolerance	RFLP	1	LCT	200 ng	1-2 weeks
Norovirus	Resistance to norovirus infections	RFLP	1	FUT2	200 ng	1-2 weeks
SNHL-1 HHL-1	Hereditary Hearing Loss	APEX	246	GJB2, GJB6, GJB3, MT-RNR1, MT-TS1, SLC26A5, SLC26A4, KCNQ4, TMC1, MYO15A, MYO7A	2 µg	4-6 weeks*
		Sequencing		GJB2	2 µg	4-6 weeks
Wilson	Wilson Disease	APEX	97	ATP7B	2 µg	4-6 weeks*

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