

# Asper Ophthalmics

Genetic test	Disease/condition name	Method	No of detectable markers	Gene	Genotyping service	Diagnostic package	
ABCA4	Stargardt Disease (STGD), Cone-Rod Dystrophy	APEX	630	ABCA4	available	available	
		Sequencing			available	available	
ADOA	Autosomal Dominant Optic Atrophy	APEX	120	OPA-1	available	available	
ADRP	Autosomal Dominant Retinitis Pigmentosa	APEX	414	Ca4, FSCN2, IMPDH1, NRL, PRPF3, PRPF31, PRPF8, RDS, RHO, ROM1, RP1, RP9, CRX, TOPORS, PNR, KLHL7	available	available	
AMD	Age-Related Macular Degeneration	Sequencing	6	ARMS2, CFH, CFB, C2	not available	available	
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	available	available	
		Sequencing		RPE65	available	available	
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	available	available	
		Sequencing		BBS1	available	available	
				BBS2	available	available	
Cornea	Corneal Dystrophy	APEX	333	COL8A2, TGFBI, VSX1, CHST6, KRT3, KRT12, GSN, TACSTD2, CYP4V2, SOD1, TCF8/ZEB1, SLC4A11, UBIAD1	available	available	
				Sequencing	BBS10	available	available
CSNB	Congenital Stationary Night Blindness	APEX	159	RHO, PDE6B, GNAT1, CABP4, GRM6, SAG, NYX, CACNA1F, CACNA2D, GRK1, TRPM1	available	available	
LCA	Leber Congenital Amaurosis	APEX	641	AIPL1, CRB1, CRX, GUCY2D, LRAT, TULP1, MERTK, RPE65, RPGRIP1, CEP290, RDH12, LCA5, SPATA7	available	available	
		Sequencing		GUCY2D	available	available	
LHON	Leber Hereditary Optic Neuropathy	RFLP	3	MT-ND4, MT-ND6, MT-ND1	available	available	
Usher	Usher Syndrome	APEX	612	CDH23, MYO7A, PCDH15, Harmonin, SANS, USH2A, VLGRI1, USH3A, Whirlin	available	available	
VMD	Vitelliform Macular Dystrophy	APEX	138	BEST1	available	available	
XLRP	X-Linked Retinitis Pigmentosa	APEX	184	RP2, RPGR	available	available	

## Asper Reprogenetics

Genetic test	Disease/condition name	Method	No of detectable markers	Gene	Genotyping service	Diagnostic package
Ashkenazi	Ashkenazi Jewish diseases testing	APEX	78	HEXA, BLM, ASPA, SMPD1, IKBKAP, DYT1, MEFV, MCOLN1, FANCC, F11, G6PC, BCKHDB, GJB2, LDL, GDE, SERPINA1, NEB, PCDH15, ABCC8, GBA, CFTR, LDLR, TMEM216	available	available
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	available	not available
CF	Cystic Fibrosis	APEX	271	CFTR	available	available
		Sequencing			available	available
CAH	Congenital Adrenal Hyperplasia	APEX, MLPA	20	CYP21A2	not available	available
Noonan	Noonan Syndrome	APEX	108	PTPN11, SOS1, RAF1, KRAS, MEK1	not available	available
NT	Syndromes related to increased Nuchal Translucency of fetus	APEX, RFLP, MLPA, Fragment Analysis	251	CYP21A2, PTPN11, SOS1, RAF1, KRAS, MEK1, DHCR7, SMN1	not available	available
SD	Skeletal Dysplasia	APEX, Sequencing	377	FGFR3, FGFR2, COL2A1, SLC26A2, ALPL, ROR2, ESCO2, CHST3, SOX9SRY	not available	available
SLO	Smith-Lemli-Opitz Syndrome	APEX	121	DHCR7	not available	available
Venous Thrombosis risk assessment	Venous Thrombosis	RFLP	4	Factor V, prothrombin/ Factor II, MTHFR	not available	available

## Asper Oncogenetics

Genetic test	Disease/condition name	Method	No of detectable markers	Gene	Genotyping service	Diagnostic package
BRAF	Colorectal Cancer, Melanoma	AS-PCR	1	BRAF	not available	available
BRCA	Hereditary Breast and Ovarian Cancer	APEX	83	BRCA1, BRCA2, CHEK2, RAD51, NBN	available	available
		Sequencing		BRCA1, BRCA2	available	available
FAP	Familial Adenomatous Polyposis	Sequencing		APC	available	available
GB	Glioblastomas	APEX	136	IDH1, IDH2, CDKN2A, EGFR, NF1, PTEN, PTPN11, RB1, TP53, TSC1, TSC2, PMS2, MLH1, MSH2, MSH6	available	not available
HNPCC/ Lynch Syndrome	Hereditary Nonpolyposis Colorectal Cancer/Lynch Syndrome	Sequencing		MLH1, MSH2, PMS2, MSH6	available	available
		MLPA		MLH1, MSH2	not available	available
MAP	MUTYH-Associated Polyposis	Sequencing		MUTYH	available	available
		RFLP	2		not available	available
MMR genes methylation analysis	Colorectal Cancer	MLPA		MLH1, MSH2, MSH6, MSH3, MLH3, PMS2	not available	available
MMR protein expression analysis	Colorectal Cancer	IHC		MLH1, MSH2, MSH6, PMS2	not available	available
MSI	Microsatellite instability	Fragment Analysis	6		not available	available
TPMT	Thiopurine S-Methyltransferase Deficiency	RFLP	3	TPMT	not available	available

Genetic test	Disease/condition name	Method	No of detectable markers	Gene	Genotyping service	Diagnostic package
ApoE	Recovery from traumatic brain injury, Alzheimer's Disease, coronary heart disease	Sequencing	2	ApoE	not available	available
Beta-Thalassemia	Beta-Thalassemia	APEX	69	HBB	available	available
Lactose Intolerance	Lactose Intolerance	RFLP	1	LCT	not available	available
Norovirus	Resistance to norovirus infections	RFLP	1	FUT2	available	not available
SNHL-1 HHL-1	Hereditary Hearing Loss	APEX	246	GJB2, GJB6, GJB3, MT-RNR1, MT-TS1, SLC26A5, SLC26A4, KCNQ4, TMC1, MYO15A, MYO7A	available	available
		Sequencing		GJB2	available	available
Wilson	Wilson Disease	APEX	97	ATP7B	available	available