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	632	ABCA4	3 µg	4-6 weeks*
		Sequencing				4-6 weeks
ADOA	Autosomal Dominant Optic Atrophy	APEX	122	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	500 ng	4-6 weeks
ARRP	Autosomal Recessive Retinitis Pigmentosa	APEX	710	CERKL, CNGA1, CNGB1, MERTK, PDE6A, PDE6B, PNR, RDH12, RGR, SAG, RLBP1, TULP1, CRB1, RP1, RPE65, USH2A, USH3A, LRAT, RBP3, PROML1, EYS, ABCA4, AIPL1, CNGA3, CNGB3, GRK1, IMPG2, RHO,	8 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	347	BBS1, BBS2, BBS3, BBS4, BBS5, BBS6, BBS7, BBS8, BBS9, BBS10, BBS11, BBS12, BBS13, PHF6, ALMS1, GNAS1	6 µg	4-6 weeks*
		Sequencing		BBS1, BBS2, BBS10	1,5 µg	4-6 weeks
Cornea	Corneal Dystrophy	APEX	333	COL8A2, TGFBI, 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, CACNA2D4, GRK1, TRPM1	3,5 µg	4-6 weeks*
LCA	Leber Congenital Amaurosis	APEX	780	AIPL1, CRB1, CRX, GUCY2D, LRAT, TULP1, MERTK, RPE65, IQCB, RPGRIP1, CEP290, RD3, RDH12, LCA5, SPATA7	6 µg	4-6 weeks*
		Sequencing		GUCY2D, RPE65		4-6 weeks
LHON	Leber Hereditary Optic Neuropathy	RFLP	3	MT-ND4, MT-ND6, MT-ND1	300 ng	4-6 weeks
Usher	Usher Syndrome	APEX	631	CDH23, MYO7A, PCDH15, USH1C, SANS, USH2A, VLGR1, USH3A, DFNB31	6 µg	4-6 weeks*
VMD	Vitelliform Macular Dystrophy	APEX	138	BEST1	1 µg	4-6 weeks*
XLRP	X-Linked Retinitis Pigmentosa	APEX	187	RP2, RPGR	2 µg	4-6 weeks*

Asper Reprogenetics

Genetic test	Disease/condition name	Method	No of detectable markers	Gene	Quantity of DNA	TAT
Ashkenazi	Ashkenazi Jewish diseases testing	APEX	108	HEXA, BLM, ASPA, SMPD1, IKBKAP, DYT1, MEFV, GBA, MCOLN1, FANCC, F11, G6PC, BCKHDB, GJB2,DLD, AGL, SERPINA1, NEB, PCDH15, ABCC8, CFTR, LDLR, TMEM216, USH3A	3,5 µg	4-6 weeks*
Carrier	Carriership testing	RFLP	94	HBB, CFTR, HEXA, HFE, F2, F5, ACADM, GALT, GJB2, MTHFR, AAT/SERPINA1, MEFV, FAH, ATP7B, SLC26A4, TCN2, DHCR7, PAH, CHRNE, FSHr	4 µg	4-6 weeks*
CF	Cystic Fibrosis	APEX	269	CFTR	2 µg	4-6 weeks*
		Sequencing				4-6 weeks
CAH	Congenital Adrenal Hyperplasia	APEX, MLPA	20	CYP21A2	1 µg	4-6 weeks
FGFR2	Apert Syndrome, Beare-Stevenson Syndrome, Craniosynostosis (nonspecific), Crouzon Syndrome, Jackson-Weiss syndrome, Pfeiffer syndrome, Saethre-Chotzen Syndrome	APEX	78	FGFR2	4,5 µg	4-6 weeks
FGFR3	Achondroplasia, Craniosynostosis/ Muenke Syndrome, Crouzon Syndrome with Acanthosis Nigricans Hypochondroplasia, Thanatophoric Dysplasia	APEX, Sequencing	43	FGFR3	4,5 µg	4-6 weeks
Noonan	Noonan Syndrome	APEX	108	PTPN11, SOS1, RAF1, KRAS, MEK1	2 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 50 ng - maternal DNA	2-3 weeks
SD	Skeletal Dysplasia	APEX, Sequencing	377	FGFR3, FGFR2, COL2A1, SLC26A2, ALPL, ROR2, ESCO2, SOX9	4,5 µg - fetal DNA 50 ng - maternal DNA	2-3 weeks
SLO	Smith-Lemli-Opitz Syndrome	APEX	121	DHCR7	500 ng	4-6 weeks
VT risk assessment	Venous Thrombosis	RFLP	4	Factor V, prothrombin/ Factor II, MTHFR	300 ng	1-2 weeks

Asper Oncogenetics

Genetic test	Disease/condition name	Method	No of detectable markers	Gene	Quantity of DNA	TAT
BRAF	Colorectal Cancer, Melanoma	AS-PCR	1	BRAF	tumor tissue	2 weeks
BRCA	Breast and Ovarian Cancer	APEX	87	BRCA1, BRCA2, CHEK2, RAD51, NBN	3 µg	4-6 weeks*
		Sequencing		BRCA1, BRCA2	3 µg	4-6 weeks
FAP	Familial Adenomatous Polyposis			APC	2 µg	4-6 weeks
GB	Glioblastoma	APEX	136	IDH1, IDH2, 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	1 µg	4-6 weeks
		Sequencing				
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

Asper Animal Genetics

Genetic test	Disease/condition name	Method	No of detectable markers	Gene	Quantity of DNA	TAT
CKCSID	Congenital Keratoconjuctivitis Sicca and Ichthyosiform Dermatosis	Sequencing	1	FAM83H	100 ng	2 weeks
EFS	Episodic Falling Syndrome	PCR	1	BCAN	100 ng	2 weeks

Varia

Genetic test	Disease/condition name	Method	No of detectable markers	Gene	Quantity of DNA	TAT
ApoE	Recovery from traumatic brain injury, Alzheimer's Disease, coronary heart disease	Sequencing	2	ApoE	300 ng	4-6 weeks
Athletic Performance	Athletic strength, speed and endurance	RFLP, PCR	2	ACE, ACTN3	300 ng	20 days
Beta-Thalassemia	Beta-Thalassemia	APEX	69	HBB	500 ng	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	249	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*

*Express delivery – The results will be delivered in 7 working days from the arrival of samples. For genotyping service only, not for diagnostic package.