

Genetic test	Disease/Conditions	Method	No of genes	No of detectable markers	Gene	Quantity of DNA	Turn Around Time*	Interpretation
ABCA4	Stargardt Disease (STGD), Cone-Rod Dystrophy	APEX	1	519	ABCA4	3 µg	3-6 weeks	yes
AD RP	Autosomal Dominant Retinitis Pigmentosa	APEX	16	385	Ca4, FSCN2, IMPDH1, NRL, PRPF3, PRPF31, PRPF8, RDS, RHO, ROM1, RP1, RP9, CRX, TOPORS, PNR, KLHL7	3 µg	3-6 weeks	yes
AR RP	Autosomal Recessive Retinitis Pigmentosa	APEX	18	585	CERKL, CNGA1, CNGB1, MERTK, PDE6A, PDE6B, PNR, RDH12, RGR, RLBP1, SAG, TULP1, CRB, RPE65, USH2A, USH3A, LRAT, PROML1	6 µg	3-6 weeks	yes
Leber-LCA	Leber Congenital Amaurosis	APEX	13	641	AIPL1, CRB1, CRX, GUCY2D, LRAT, TULP1, MERTK, RPE65, RPGRIP1, CEP290, RDH12, LCA5, SPAYTA7	6 µg	3-6 weeks	yes

BBS	Bardet-Biedl Syndrome	APEX	14	308	BBS1, BBS2, BBS3, BBS4, BBS5, BBS6, BBS7, BBS8, BBS9, BBS10, BBS12, PHF6, ALMS1, GNAS1	5 µg	3-6 weeks	yes
BEST-VMD	Vitelliform Macular Dystrophy	APEX	1	138	BEST1	1 µg	3-6 weeks	yes
CSNB	Congenital Stationary Night Blindness	APEX	9	126	RHO, PDE6B, GNAT1, CABP4, GRM6, SAG, NYX, CACNA1F, CACNA2D	2,5 µg	3-6 weeks	yes
Cornea	Corneal Dystrophy	APEX	13	325	COL8A2, TGFB1, VSX1, CHST6, KRT3, KRT12, GSN, TACSTD2, CYP4V2, SOD1, TCF8/ZEB1, SLC4A11, UBIAD1	3 µg	3-6 weeks	yes
OPA-1 ADOA	Autosomal Dominant Optic Atrophy	APEX	1	118	OPA-1	2 µg	3-6 weeks	yes
AMD	Age-Related Macular Degeneration	Cycle Sequencing	2	3	ARMS2, CFH	200 ng	3-6 weeks	yes
Usher	Usher Syndrome	APEX	9	612	CDH23, MYO7A, PCDH15, Harmonin, SANS, Usherin, VLGR1, USH3A,	6 µg	3-6 weeks	yes



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**\* Express delivery – The results will be delivered in 3–5 working days from the arrival of samples.**