

S.No	OPHTHAMOLOGY	Gene list	Specimen Type	Methodology	TAT
1	LHON hotspot mutation testing		Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	14 days
2	Comprehensive Ophthalmic genetic Disorder Panel	TBK1, PLA2G5, GZF1, COL18A1, WRN, TMEM107, PRICKLE3, DNMT1, IKBKG, WDR19, RPE65, MFRP, ESCO2, AIPL1, ZNF64, SEC23A, MYF5, FOXC1, ARR3, TEK, FOXA2, PDE6H, HSPG2, CERKL, IFT80, TPP1, PGK1, JAM3, CNGA3, YME1L1, POLG2, MIP1, POMGNT1, APOA1, COL9A3, CRPPA, RGS9, NEU1, ELOVL4, GPR143, BBIP1, PEX16, SALL2, CLDN19, FBLN5, LARGE1, TRPM1, PITPNM3, KIAA1549, COL11A1, TIMM50, UNC45B, MVK, PRDM13, ARMCS9, CREBBP, FNBP4, RP1, NPHP3, ERCC5, BBS7, YWHAE, SDHA, OTX2, FLNA, PEX19, CCDC28B, TCTN3, POLG, HMGB3, COL9A1, B3GALT1, TMEM98, PRPF4, ITM2B, CRYGB, SALL1, TUBGCP4, FAM161A, LCA5, ALX1, C	Peripheral Blood in EDTA (Purple Top), DIRECT DNA	Next Generation Sequencing (NGS)	21 days
3	Glaucoma gene panel	LMX1B, OPTN, TEK, CYP1B1, GLIS3, SBF2, COL18A1, MYOC, FOXE3, PITX3, BEST1, LTBP2, FOXC1, PAX6, ASB10	Peripheral Blood in EDTA (Purple Top), DIRECT DNA	Next Generation Sequencing (NGS)	21 days
4	Duane retraction syndrome (CHN1 and MAFB gene sequencing)	MAFB, CHN1	DIRECT DNA, Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
5	PRPH2 and GUCY2D gene sequencing	PRPH2, GUCY2D	DIRECT DNA, Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
6	CHM gene sequencing	CHM	DIRECT DNA, Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
7	GPR143 and FRMD7 gene sequencing	GPR143, FRMD7	Peripheral Blood in EDTA (Purple Top), DIRECT DNA	Next Generation Sequencing (NGS)	19 days
8	CYP1B1 gene sequencing		DIRECT DNA, Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
9	TULP1 gene sequencing		Peripheral Blood in EDTA (Purple Top), DIRECT DNA	Next Generation Sequencing (NGS)	19 days
10	WAGR syndrome (PAX6 and WT1) gene sequencing		Peripheral Blood in EDTA (Purple Top), DIRECT DNA	Next Generation Sequencing (NGS)	19 days
11	RPGR gene sequencing		Peripheral Blood in EDTA (Purple Top), DIRECT DNA	Next Generation Sequencing (NGS)	19 days
12	PRSS56 and MFRP gene analysis		Peripheral Blood in EDTA (Purple Top), DIRECT DNA	Next Generation Sequencing (NGS)	19 days
13	PROM1 gene analysis		Peripheral Blood in EDTA (Purple Top), DIRECT DNA	Next Generation Sequencing (NGS)	19 days

14	Usher syndrome gene panel	CDH23,PCDH15,PDZD7,CIB2,USH1C,CLRN1,WHRN,USH1G,USH2A,HARS1,ADGRV1,MYO7A	Peripheral Blood in EDTA (Purple Top),DIRECT DNA,Amniotic fluid (AF),Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	19 days
15	Retinal Degeneration gene panel		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	19 days
16	Optic atrophy gene panel		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	19 days
17	Microphthalmia and anophthalmia gene panel		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
18	Leber congenital amaurosis gene panel		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
19	Congenital cataract gene panel	INPP5K,WFS1,VIM,CRYGS,LIM2,CRYGD,LEMD2,FTL,P3H2,NHS,CRYAB,GALK1,FYCO1,PITX3,CRYGB,HSF4,HMX1,UNC45B,TMEM114,CRYGC,CHMP4B,CDK5RAP2,FOXO3,FAM126A,MOMO1,CRYAA,SLC33A1,CRYBB3,CRYBB2,GJA8,GJA3,TDRD7,EPA2,MAF,LSS,BFSP2,BFSP1,EYA1,MIP,GCNT2,SLC16A12,GALT,SIL1,CRYBB1,CRYBA4,GFER,CTDP1,AGK,CRYBA1,RDH11,CRYBA2	Peripheral Blood in EDTA (Purple Top),DIRECT DNA,Amniotic fluid (AF),Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	19 days
20	Choroideremia (CHM) deletion/duplication analysis		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	MLPA	14 days
21	C8orf37 gene sequencing		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	19 days
22	Corneal dystrophy gene panel		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	19 days

23	Usher syndrome (USH2A) deletion/duplication analysis		Peripheral Blood in EDTA (Purple Top),Product of conception (POC),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	MLPA	14 days
24	Congenital stationary night blindness gene panel		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	19 days