Phosphorus Diagnostics

Ophthalmology Panel List

Description	# of Genes	Genes	ICD-10 Suggestions*
RETINAL DISORDERS			
Pan Inherited Retinal Disorders			
Includes the genes most commonly associated with inherited retinal disorders.	144	ABCA4 ABHD12 ADAM9 ADGRV1 AIPL1 ARL6 ATF6 BBS1 BBS10 BBS12 BBS2 BBS4 BBS5 BBS7 BBS9 BEST1 C2ORF71 C8ORF37 CA4 CABP4 CACNA1F CACNA2D4 CAPN5 CDH23 CDH3 CDHR1 CEP290 CERKL CIB2 CLRN1 CNGA1 CNGA3 CNGB1 CNGB3 CNNM4 COL11A1 COL2A1 COL9A1 CRB1 CRX CYP4V2 DHDDS EFEMP1 ELOVL4 EMC1 EYS FAM161A FBLN5 FLVCR1 FZD4 GDF6 GNAT1 GNAT2 GPR179 GRK1 GRM6 GUCA1A GUCY2D HARS HGSNAT HK1 IDH3B IFT140 IFT172 IMPDH1 IMPG1 IMPG2 IQCB1 KCNJ13 KCNV2 KIZ KLHL7 LCA5 LRAT LRIT3 LZTFL1 MAK MERTK MKKS MKS1 MVK MYO7A NDP NMNAT1 NPHP1 NPHP4 NR2E3 NRL NYX OAT OPA3 OTX2 PCDH15 PDE6A PDE6B PDE6C PDE6G PDE6H PDZD7 PRCD PROM1 PRPF3 PRPF31 PRPF6 PRPF8 PRPH2 RAX2 RBP3 RBP4 RD3 RDH12 RDH5 RGR RHO RLBP1 RP1 RP1L1 RP2 RPE65 RPGR RPGRIP1 RS1 SAG SDCCAG8 SEMA4A SNRNP200 SPATA7 TIMP3 TMEM126A TOPORS TRIM32 TRNT1 TRPM1 TSPAN12 TTC8 TULP1 USH1C USH1G USH2A VCAN WDPCP WHRN ZNF513	H35.50 Hereditary retinal dystrophy H53.6 Congenital stationary night blindness H53.5 Achromatopsia Q87.89 Bardet-Biedl syndrome H54.0 Blindness Z84.81 Family history of carrier of genetic disease
Emerging Evidence Genes			
Add-on genes for which there is initial evidence that mutations in these genes may be related to inherited retinal disorders.	17	DHX38 DTHD1 FSCN2 GUCA1B INPP5E KIAA1549 NEK2 PITPNM3 PRDM13 RIMS1 ROM1 RP9 SLC24A1 SLC7A14 SPP2 UNC119 ZNF408	
Achromatopsia			
Includes the genes most commonly associated with achromatopsia.	6	ATF6 CNGB3 CNGA3 GNAT2 PDE6C PDE6H	H53.5 Achromatopsia Z84.81 Family history of carrier of genetic disease
Bardet-Biedl Syndrome			
Includes the genes most commonly associated with Bardet-Biedl syndrome.	17	ARL6 BBS1 BBS10 BBS12 BBS2 BBS4 BBS5 BBS7 BBS9 CEP290 LZTFL1 MKKS MKS1 SDCCAG8 TRIM32 TTC8 WDPCP	Q87.89 Bardet-Biedl syndrome H35.52 Pigmentary Retinal Dystrophy Z84.81 Family history of carrier of genetic disease
Emerging Evidence Genes			
Add-on genes for which there is initial evidence that mutations in these genes may be related to Bardet-Biedl syndrome.	2	IFT172 INPP5E	
Cone-Rod Dystrophy			
Includes the genes most commonly associated with cone-rod dystrophy.	26	ABCA4 ADAM9 BEST1 C8ORF37 CABP4 CACNA1F CACNA2D4 CDHR1 CDHR1 CERKL CNGB3 CNNM4 CRX CYP4V2 GUCA1A GUCY2D KCNV2 MERTK PDE6C PDE6H PROM1 PRPH2 RAX2 RDH5 RPGR RPRIP1 SEMA4A	H35.50 Hereditary retinal dystrophy H54.0 Blindness Z84.81 Family history of carrier of genetic disease
Emerging Evidence Genes			
Add-on genes for which there is initial evidence that mutations in these genes may be related to cone-rod dystrophy.	4	AIPL1 PITPNM3 RIMS1 UNC119	

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Description	# of Genes	Genes	ICD-10 Suggestions*
RETINAL DISORDERS, CONTINUED			
Congenital Stationary Night Blindness			
Includes the genes most commonly associated with congenital stationary night blindness.	16	CABP4 CACNA1F CYP4V2 GNAT1 GPR179 GPR179 GRK1 GRM6 LRIT3 NYX PDE6B RDH5 RHO RLBP1 RPE65 SAG TRPM1	H53.6 Congenital stationary night blindness H54.0 Blindness Z84.81 Family history of carrier of genetic disease
Emerging Evidence Genes			
Add-on genes for which there is initial evidence that mutations in these genes may be related to congenital stationary night blindness.	2	CACNA2D4 SLC24A1	
Leber Congenital Amaurosis			
Includes the genes most commonly associated with Leber congenital amaurosis.	21	AIPL1 CABP4 CEP290 CRB1 CRX GDF6 GUCY2D IQCB1 KCNJ13 LCA5 LRAT MERTK NMNAT1 OTX2 PRPH2 RD3 RDH12 RPE65 RPRIP1 SPATA7 TULP1	H35.50 Hereditary retinal dystrophy H54.0 Blindness Z84.81 Family history of carrier of genetic disease
Emerging Evidence Genes			
Add-on genes for which there is initial evidence that mutations in these genes may be related to Leber congenital amaurosis.	4	BBS4 DTHD1 IMPDH1 SNRNP200	
Macular Dystrophy			
Includes the genes most commonly associated with macular dystrophy.	21	ABCA4 BEST1 CDH3 CERKL CNGB3 CRB1 EFEMP1 ELOVL4 FBLN5 IMPG1 PROM1 PRPH2 RAX2 RBP4 RDH12 RDH5 RLBP1 RP1L1 RPGR RS1 TIMP3	H35.50 Hereditary retinal dystrophy H35.30 Macular Degeneration H54.0 Blindness Z84.81 Family history of carrier of genetic disease
Emerging Evidence Genes			
Add-on genes for which there is initial evidence that mutations in these genes may be related to macular dystrophy.	3	FSCN2 GUCA1B PRDM13	
Retinitis Pigmentosa			
Includes the genes most commonly associated with retinitis pigmentosa.	72	ABCA4 ABHD12 ARL6 BBS1 BBS2 BEST1 C2ORF71 C8ORF37 CA4 CDHR1 CEP290 CERKL CLRN1 CNGA1 CNGB1 CRB1 CRX CYP4V2 DHDDS EMC1 EYS FAM161A FLVCR1 GUCY2D HGSNAT HK1 IDH3B IFT140 IFT172 IMPDH1 IMPG2 KIZ KLHL7 LCA5 LRAT MAK MERTK MVK NMNAT1 NR2E3 NRL PDE6A PDE6B PDE6G PRCD PROM1 PRPF3 PRPF31 PRPF6 PRPF8 PRPH2 RBP3 RDH12 RGR RHO RLBP1 RP1 RP2 RPE65 RPGR RPGRIP1 SAG SEMA4A SNRNP200 SPATA7 TOPORS TRNT1 TTC8 TULP1 USH2A WDR19 ZNF513	H35.50 Hereditary retinal dystrophy H35.52 Pigmentary Retinal Dystrophy H54.0 Blindness Z84.81 Family history of carrier of genetic disease
Emerging Evidence Genes			
Add-on genes for which there is initial evidence that mutations in these genes may be related to retinitis pigmentosa.	11	AIPL1 DHX38 FSCN2 GUCA1B KIAA1549 NEK2 ROM1 RP9 SLC7A14 ZNF408	
Usher Syndrome			
Includes the genes most commonly associated with Usher syndrome.	13	ABHD12 ADGRV1 CDH23 CIB2 CLRN1 HARS MYO7A PCDH15 PDZD7 USH1C USH1G USH2A WHRN	H35.50 Hereditary retinal dystrophy H54.0 Blindness Z84.81 Family history of carrier of genetic disease

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Description	# of Genes	Genes	ICD-10 Suggestions*
CATARACTS			
Congenital Cataracts			
Includes the genes most commonly associated with hereditary colon cancer, including both polyposis syndromes and non-polyposis syndromes.	38	AGK BCOR BFSP1 BFSP2 CRYAA CRYAB CRYBA1 CRYBB1 CRYBB2 CRYBB3 CRYGC CRYGD CRYGS CTDP1 EPHA2 EYA1 FAM126A FOXC1 FOXE3 FYCO1 GALK1 GCNT2 GJA3 GJA8 HSF4 MAF MIP MIR184 NHS OCRL P3H2 PAX6 PITX2 PITX3 PXDN SIL1 SLC33A1 VSX2	Q12.0 Congenital Cataract Z84.81 Family history of carrier of genetic disease
Emerging Evidence Genes			
Add-on genes for which there is initial evidence that mutations in these genes may be related to hereditary colorectal cancer.	6	CHMP4B CRYBA4 CRYGB LIM2 TDRD7 VIM	
GLAUCOMA			
Early-Onset Glaucoma			
Includes the genes most commonly associated with early-onset glaucoma.	12	COL4A1 CYP1B1 FOXC1 LMX1B LTBP2 MFRP MYOC OPTN PAX6 PITX2 SH3PXD2B WDR36	Q15.0 Congenital Glaucoma H40.1 Open-Angle Glaucoma Z84.81 Family history of carrier of genetic disease
CORNEAL DYSTROPHY			
Corneal Dystrophy			
Includes genes associated with corneal dystrophy, including TGFBI-related disorders that cause risk of vision loss in individuals undergoing laser surgery.	8	CYP4V2 FOXE3 GJA8 LCAT MAF PITX2 TGFBI VSXI	H18.50 Hereditary Corneal Dystrophy Z84.81 Family history of carrier of genetic disease

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