**پانل بیماری های چشمی**

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| **ردیف** | **نام بیماری** | **تعداد ژن** | **ژن های مورد بررسی** |
| 1 | Usher Syndrome | 15 | ABHD12, CDH23, CIB2, CLRN1, DFNB31, GPR98, HARS, MYO7A, PCDH15, PDZD7, PEX1, PEX6, USH1C, USH1G, USH2A |
| 2 | Retinitis Pigmantosa  (autosomal dominant and X-linked) | 30 | ARL3, BEST1, CA4, CRX, FSCN2, GUCA1B, HK1, IMPDH1, KLHL7, NR2E3, NRL, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RDH12, RGR, RHO, ROM1, RP1, RP2, RP9, RPE65, RPGR, SEMA4A, SNRNP200, SPP2, TOPORS |
| 3 | Retinitis Pigmantosa  (autosomal recessive and X-linked) | 63 | ABCA4, ARL2BP, BBS1, BBS2, BEST1, C2orf71, C8orf37, CDHR1, CERKL, CLRN1, CNGA1, CNGB1, CRB1, CYP4V2, DHDDS, DHX38, EMC1, EYS, FAM161A, FLVCR1, GNPTG, GPR125, HGSNAT, IDH3B, IFT172, IMPG2, KIAA1549, KIZ, LRAT, MAK, MERTK, NEK2, NMNAT1, NR2E3, NRL, PDE6A, PDE6B, PDE6G, PRCD, PROM1, PRPF31, RBP3, RBP4, RDH11, RDH12, RGR, RHO, RLBP1, RP1, RP1L1, RP2, RPE65, RPGR, SAG, SLC7A14, SPATA7, TRNT1, TTC8, TUB, TULP1, USH2A, ZNF408 , ZNF513 |
| 4 | Achromatopsia | 6 | CNGB3 (Ex. 10), 2.) ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H |
| 5 | Bardet-Biedl Syndrome | 24 | ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CCDC28B, CEP290, IFT172, IFT27, LZTFL1, MKKS, MKS1, NPHP1, SDCCAG8, TRIM32, TTC8, TTC21B, WDPCP |
| 6 | Congenital Stationary Night Blindness | 15 | CABP4, CACNA1F, CACNA2D4, GNAT1, GPR179, GRK1, GRM6, LRIT3, NYX, PDE6B, RBP4, RHO, SAG, SLC24A1, TRPM1 |
| 7 | Joubert Syndrome | 28 | AHI1, ARL13B, B9D1, C5orf42, CC2D2A, CEP104, CEP164, CEP290, CEP41, CSPP1, INPP5E, KIAA0556, KIAA0586, KIF7, NPHP1, OFD1, PDE6D, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423 |
| 8 | Leber Congenital Amaurosis | 21 | AIPL1, CEP290, CRB1, CRX, GUCY2D, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, MERTK, NMNAT1, OTX2, PRPH2, RD3, RDH12, RDH5, RPE65, RPGRIP1, SPATA7, TULP1 |
| 9 | Refsum Disease | 7 | PEX1, PEX2, PEX26, PEX3, PEX5, PEX7, PHYH |
| 10 | Senior Loken Syndrome | 12 | CEP164, CEP290, IFT81, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19, ZNF423 |
| 11 | Stargardt Disease and Macular Dystrophies | 23 | ABCA4, BEST1, C1QTNF5, CDH3, CLN3, CNGB3, CRB1, CRX, CTNNA1, DRAM2, ELOVL4, FSCN2, IMPG1, IMPG2, MFSD8, PRDM13, PROM1, PRPH2, RDH12, RP1L1, RPGR, TIMP3, TTLL5 |
| 12 | Cone Rod Dystrophies | 40 | ABCA4, ACBD5, ADAM9, AIPL1, C21orf2, C2orf71, C8orf37, CABP4, CACNA1F, CACNA2D4, CDHR1, CEP290, CERKL, CNGA3, CNGB3, CNNM4, CRB1, CRX, CYP4V2, GNAT2, GUCA1A, GUCY2D, KCNV2, PCYT1A, PDE6C, PDE6H, PITPNM3, POC1B, PROM1, PRPH2, RAB28, RAX2, RDH5, RGS9, RGS9BP, RIMS1, RPGR, RPGRIP1, SEMA4A, TTLL5 |
| 13 | Flecked Retina Disorders | 7 | CHM, EFEMP1, PLA2G5, RDH5, RLBP1, RS1, VPS13B |
| 14 | Familial Exudative Vitreoretinopathy and Wagner Syndrome | 8 | CAPN5, COL2A1, FZD4, LRP5, NDP, TSPAN12, VCAN, ZNF408 |
| 15 | Stickler Syndrome | 5 | COL11A1, COL11A2, COL2A1, COL9A1, COL9A2 |
| 16 | Optic Atrophy | 15 | ACO2, AFG3L2, C12orf65, CISD2, MFN2, NR2F1, OPA1, OPA3, RTN4IP1, SLC25A46, SPG7, TIMM8A, TMEM126A, VAX2, WFS1  incl. LHON (Leber Hereditary Optic Neuropathy) optional – MT-ND1, m.3460G>A; MT-ND4, m.11778G>A; MT-ND6, m.14484T>C |
| 17 | Albinism | 9 | C10orf11, GPR143, LYST, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1 |
| 18 | Hermansky-Pudlak Syndrome | 9 | AP3B1, BLOC1S3, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6 |
| 19 | Microphthalmia | 26 | ABCB6, ALDH1A3, BCOR, BMP4, CHD7, FREM1, GDF3, GDF6, HCCS, HMGB3, MAB21L2, MFRP, NAA10, OTX2, PRSS56, RARB, RAX, RBP4, SHH, SIX6, SMOC1, SOX2, STRA6, TENM3, VAX1, VSX2 |
| 20 | Cataract | 41 | AGK, BCOR, BFSP1, BFSP2, CHMP4B, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGA, CRYGB, CRYGC, CRYGD, CRYGS, EPHA2, EYA1, FOXE3, FTL, FYCO1, GALK1, GCNT2, GJA3, GJA8, HSF4, LEPREL1, LIM2, LSS, MAF, MIP, NHS, PAX6, PITX3, PXDN, SIL1, SIPA1L3, SLC16A12, TDRD7, VIM |
| 21 | Septo-optical Dysplasia (SOD) | 7 | FGFR2, HESX1, OTX2, PROKR2, SOX2, SOX3, TAX1BP3 |
| 22 | Complete Panel – Eye Disease | 287 | ABCA4, ABCB6, ABHD12, ACBD5, ADAM9, AGK, AHI1, AIPL1, ALDH1A3, ALMS1, AP3B1, ARL13B, ARL2BP, ARL6, ATF6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BEST1, BFSP1, BFSP2, BLOC1S3, BLOC1S6, BMP4, C10orf11, C1QTNF5, C21orf2, C2orf71, C5orf42, C8orf37, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CCDC28B, CDH23, CDH3, CDHR1, CEP164, CEP290, CEP41, CERKL, CHD7, CHM, CHMP4B, CIB2, CLN3, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, CRB1, CRX, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGA, CRYGC, CRYGD, CRYGS, CSPP1, CYP4V2, DFNB31, DHDDS, DHX38, DTNBP1, EFEMP1, ELOVL4, EMC1, EPHA2, EYA1, EYS, FAM161A, FGFR2, FLVCR1, FOXE3, FREM1, FSCN2, FTL, FYCO1, FZD4, GALK1, GCNT2, GDF3, GDF6, GJA3, GJA8, GNAT1, GNAT2, GNPTG, GPR125, GPR143, GPR179, GPR98, GRK1, GRM6, GUCA1A, GUCA1B, GUCY2D, HARS, HCCS, HESX1, HK1, HMGB3, HPS1, HPS3, HPS4, HPS5, HPS6, HSF4, IDH3B, IFT172, IFT27, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, KCNJ13, KCNV2, KIAA1549, KIF7, KIZ, KLHL7, LCA5, LEPREL1, LIM2, LRAT, LRIT3, LRP5, LYST, LZTFL1, MAB21L2, MAF, MAK, MC1R, MERTK, MFN2, MFRP, MIP, MKKS, MKS1, MYO7A, NAA10, NDP, NEK2, NHS, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NYX, OCA2, OFD1, OPA1, OPA3, OTX2, PAX6, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6D, PDE6G, PDE6H, PDZD7, PEX1, PEX2, PEX26, PEX7, PHYH, PITPNM3, PITX3, PLA2G5, PRCD, PROKR2, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRSS56, PXDN, RAB28, RARB, RAX, RAX2, RBP3, RBP4, RD3, RDH11, RDH12, RDH5, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, SAG, SDCCAG8, SEMA4A, SHH, SIL1, SIX6, SLC16A12, SLC24A1, SLC45A2, SLC7A14, SMOC1, SNRNP200, SOX2, SOX3, SPATA7, SPG7, STRA6, TCTN1, TCTN2, TCTN3, TDRD7, TENM3, TIMP3, TMEM126A, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TRIM32, TRPM1, TSPAN12, TTC21B, TTC8, TTLL5, TUB, TULP1, TYR, TYRP1, USH1C, USH1G, USH2A, VAX1, VCAN, VIM, VPS13B, VSX2, WDPCP, ZNF423, ZNF513 |