



Eye Disorders Gene List
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سپاهی خاص
شماره ثبت: ۴۱۰۴۵

تاریخ:
شماره:
پیوست:

ABCA1	CFD	GUCA1B	OCA2	RPGR
ABCA4	CFH	GUCY2D	OFD1	RPGRIP1
ACE	CFHR1	HCCS	OPA1	RPGRIP1L
ADIPOR1	CFHR3	HMCN1	OPA3	RS1
AHI1	CFI	HPS1	OTX2	SAG
AIPL1	CHST6	HPS3	PAX6	SDCCAG8
AP3B1	CLRN1	HPS4	PCDH15	SEMA4A
APOE	CNGA1	HPS5	PDE6A	SERPING1
ARL13B	CNGA3	HPS6	PDE6B	SHH
ARL6	CNGB1	HTRA1	PDE6C	SIX6
ARMS2	CNGB3	IDH3B	PDE6G	SLC24A1
BBS1	COL1A2	IGFBP7	PDE6H	SLC45A2
BBS10	COL2A1	IMPDH1	PDZD7	SNRNP200
BBS12	CRB1	IMPG2	PIKFYVE	SOX2
BBS2	CRP	INPP5E	PITPNM3	SPATA7
BBS4	CRX	IQCB1	PLEKHA1	STRA6
BBS5	CRYBA4	KCNJ13	POLR2B	TEAD1
BBS7	CST3	KCNV2	PON1	TGFBI
BBS9	CX3CR1	KIF21A	PRCD	TIMP3
BCOR	CYP4V2	KIF7	PROM1	TLR3
BEST1	DCN	KLHL7	PRPF3	TLR4
BLOC1S3	DFNB31	KRT12	PRPF31	TMEM126A
BLOC1S6	DHDDS	KRT3	PRPF6	TMEM216
BMP4	DTNBP1	LCA5	PRPF8	TMEM67
C1QTNF5	EFEMP1	LIPC	PRPH2	TNFRSF10A
C2	ELOVL4	LPL	RAX	TOPORS
C2orf71	ERCC6	LRAT	RAX2	TRIM32
C3	ESR1	LRP5	RD3	TRPM1
C4orf14	EYS	LRP6	RDH12	TSPAN12
C9	FAM161A	MAK	RDH5	TTC8
CA4	FBLN5	MERTK	REST	TUBB3
CABP4	FLVCR1	MITF	RGR	TULP1
CACNA1F	FRMD7	MKKS	RHO	TYR
CACNA2D4	FSCN2	MKS1	RIMS1	TYRP1
CC2D2A	FZD4	MYO7A	RLBP1	UBIAD1
CCR2	GDF6	NDP	ROBO1	UNC119
CDH23	GNAT1	NMNAT1	ROM1	USH1C
CDH3	GNAT2	NPHP1	RORA	USH1G
CDHR1	GPR143	NPHP4	RP1	USH2A
CEP290	GPR98	NR2E3	RP1L1	VEGFA
CERKL	GRK1	NRL	RP2	VLDLR
CETP	GRM6	NYX	RP9	VSX1
CFB	GUCA1A	OAT	RPE65	ZNF513



Important Notes:

- 1- Only known exons of these genes will be examined
- 2- Repeat expansion disorders will not be covered
- 3- Genomic regions beside exons of protein-coding genes, genes that are not listed here, repeat expansions and mutations in the upstream and downstream regulatory regions will not be investigated.

Additional Comments:

- Although next generation sequencing (NGS) is a method of choice for high throughput sequencing purposes, **NGS has not been approved for clinical and diagnostic use**; therefore, Sanger sequencing must be done to confirm the sequencing data, particularly on identified mutations.
- Genetic counseling is recommended to explain risks and potential pitfalls of the experiment.
- It is of utmost importance for all clinicians involved in the care of families requesting molecular genetic diagnostic tests and the families themselves to be aware of the risk of errors in DNA analysis. Incorrect analysis may result from 1) incorrect data and clinical diagnosis 2) incomplete family studies and history 3) mix-up of DNA samples and mislabeling 4) rare molecular events 5) new or spontaneous mutations 6) paternity problems, adaptation, IVF, egg donor, bone marrow transplantation, recent blood product transfusion 7) maternal DNA contamination of CVS or amniotic fluid samples 8) technical errors. The risk of errors from various reasons mentioned above and several others is about 0.5%, while the chance of technical errors of all types is estimated to be around 0.5%. The risk of errors due to DNA recombination in diagnosis is approximately 0.3%. We take no responsibility about patient identity and possible mis-labeling of the DAN samples. Any feedback from our colleagues in the clinical field would be most welcomed. Comments can be given in writing or by calling my number listed below or by e-mail to:
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