DIVAs report

GENERAL INFORMATION

Date	2023-10-12
DIVAs version	DIVAs v1.1

HPO TERMS

HPO TERM	DESCRIPTION
HP:0000662	Nyctalopia
HP:0000505	Visual impairment
HP:0007703	Abnormality of retinal pigmentation
HP:0007947	Pericentral retinitis pigmentosa
HP:0000546	Retinal degeneration

FAMILY HISTORY

Sample	Father	Mother	Sex	Affected
				Yes

ANALYSIS PARAMETERS

QUALITY	30.0	Only variants with QUALITY >= of this threshold will be retained
GENOTYPE QUALITY	0.0	Only variants with GENOTYPE QUALITY >= of this threshold will be retained
FILTER	PASS	Only variants with FILTER field with this value will be retained
PATHOGENICITY SCORE	1.0	Only variants with PS >= of this threshold will be retained

STATISTICS

TOTAL VARIANTS	229267	Variants present in the original VCF
RETAINED VARIANTS	1899	Variants with QUALITY >= 30.0, GENOTYPE QUALITY >= 0.0, FILTER=PASS and PS >= 1.0
TOTAL COMBINATIONS	111639	Number of digenic combinations evaluated by DIVAs
NUMBER OF PATHOGENIC COMBINATIONS	2859	Number of digenic combinations classified pathogenic by DIVAs

RESULTS

Gene A	HGVS.c A	HGVS.p A	Effect A	Gene B	HGVS.c B	HGVS.p B	Effect B	Score
TRPM1	c.1879G>A	p.Val627Met	missense	RP1	c.1625C>G	p.Ser542*	stop gained	1.00000
RBP3	c.2919C>A	p.Ser973Ser	synonymous	RP1	c.1625C>G	p.Ser542*	stop gained	1.00000
CNGB1	c.874+2074G>T; c.291-145A>C	.; .	intron; intron	RP1	c.1625C>G	p.Ser542*	stop gained	0.99999
VCAN	c.*386G>A	.	3 prime UTR	RP1	c.1625C>G	p.Ser542*	stop gained	0.99999
BEST1	c.*1721G>A; c.*1692G>A	.; .	3 prime UTR; 3 prime UTR	RP1	c.1625C>G	p.Ser542*	stop gained	0.99999
PRPH2	c.582-4734_582-4733insGTTT	.	intron	RP1	c.1625C>G	p.Ser542*	stop gained	0.99999
PROM1	c.1683-1269_1683-1267delGTC; c.1683-1283_1683-1275delCCTTCTTCT	.; .	intron; intron	RP1	c.1625C>G	p.Ser542*	stop gained	0.99998
RIMS1	c.904G>C	p.Glu302Gln	missense	RP1	c.1625C>G	p.Ser542*	stop gained	0.99998
VCAN	c.*386G>A	.	3 prime UTR	TRPM1	c.1879G>A	p.Val627Met	missense	0.99998
TULP1	c.823-754_823-750delTACAC	.	intron	RP1	c.1625C>G	p.Ser542*	stop gained	0.99998
CHM	c.1244+97A>T	.	intron	RP1	c.1625C>G	p.Ser542*	stop gained	0.99997

ERCC3	c.1945+1915_1945+1918delAAAA	.	intron	RP1	c.1625C>G	p.Ser542*	stop gained	0.99997
POC1B	c.1114-12674_1114-12672delTTT	.	intron	RP1	c.1625C>G	p.Ser542*	stop gained	0.99996
AHR	c.2404-73T>C	.	intron	RP1	c.1625C>G	p.Ser542*	stop gained	0.99993
GPR143	c.250+1044_250+1063delTGTGTGTGTGTGTGTG	.	intron	RP1	c.1625C>G	p.Ser542*	stop gained	0.99993
ZNF141	c.204_205insGA; c.209_210delTC	p.Lys69fs; p.Ile70fs	frameshift; frameshift	RP1	c.1625C>G	p.Ser542*	stop gained	0.99992
ERCC4	c.1812-541G>A	.	intron	RP1	c.1625C>G	p.Ser542*	stop gained	0.99989
VCAN	c.*386G>A	.	3 prime UTR	BEST1	c.*1721G>A; c.*1692G>A	.; .	3 prime UTR; 3 prime UTR	0.99988
PRIMPOL	c.-59-7delG	.	splice region, intron	RP1	c.1625C>G	p.Ser542*	stop gained	0.99988
POLR3A	c.3472C>T	p.Leu1158Phe	missense	RP1	c.1625C>G	p.Ser542*	stop gained	0.99986
DHX38	c.3477+5G>A; c.*246T>G	.; .	splice region, intron; 3 prime UTR	RP1	c.1625C>G	p.Ser542*	stop gained	0.99983
LPL	c.344C>A; c.875G>A	p.Ser115*; p.Ser292Asn	stop gained; missense	RP1	c.1625C>G	p.Ser542*	stop gained	0.99980
FTH1	c.541_543delAAT; c.388-1_388insATCCCCAC	p.Asn181del; .	conservative inframe deletion; splice acceptor, intron	RP1	c.1625C>G	p.Ser542*	stop gained	0.99979

DHX37	c.2045+1648T>C	.	intron	RP1	c.1625C>G	p.Ser542*	stop gained	0.99978
BEST1	c.*1721G>A; c.*1692G>A	.; .	3 prime UTR; 3 prime UTR	RBP3	c.2919C>A	p.Ser973Ser	synonymous	0.99977
GRIK2	c.899G>A	p.Arg300Gln	missense	RP1	c.1625C>G	p.Ser542*	stop gained	0.99976
SDCCAG8	c.1744+124C>T	.	intron	RP1	c.1625C>G	p.Ser542*	stop gained	0.99976
RIMS1	c.904G>C	p.Glu302Gln	missense	POC1B	c.1114-12674_1114-12672delTTT	.	intron	0.99976
RIMS1	c.904G>C	p.Glu302Gln	missense	TRPM1	c.1879G>A	p.Val627Met	missense	0.99974
COL3A1	c.4011+1G>T; c.3997G>A	.; p.Asp1333Asn	splice donor, intron; missense	RP1	c.1625C>G	p.Ser542*	stop gained	0.99963
CNBP	c.*458C>A	.	3 prime UTR	RP1	c.1625C>G	p.Ser542*	stop gained	0.99960
HTT	c.102delG; c.99_100delGC	p.Gln34fs; p.Gln34fs	frameshift; frameshift	RP1	c.1625C>G	p.Ser542*	stop gained	0.99955
POC1B	c.1114-12674_1114-12672delTTT	.	intron	TRPM1	c.1879G>A	p.Val627Met	missense	0.99955
ARMC2	c.1024-3233G>A	.	intron	RP1	c.1625C>G	p.Ser542*	stop gained	0.99954
CPE	c.41G>C; c.1113_1113+1insATACACCGAGGTGTTAAAGGGTTTGTCCGTGACCTTCAGG	p.Gly14Ala; .	missense; splice donor, intron	RP1	c.1625C>G	p.Ser542*	stop gained	0.99954
DNAAF4	c.862_866delAAGAA	p.Lys288fs	frameshift	RP1	c.1625C>G	p.Ser542*	stop gained	0.99951
TULP1	c.823-754_823-750delTACAC	.	intron	BEST1	c.*1721G>A; c.*1692G>A	.; .	3 prime UTR; 3 prime UTR	0.99950

RB1	c.607+2970T>C	.	intron	RP1	c.1625C>G	p.Ser542*	stop gained	0.99950
BBS9	c.2116-30675dupA	.	intron	RP1	c.1625C>G	p.Ser542*	stop gained	0.99947
CDH2	c.*804T>A	.	3 prime UTR	RP1	c.1625C>G	p.Ser542*	stop gained	0.99946
DDX3X	.	.	intron	RP1	c.1625C>G	p.Ser542*	stop gained	0.99946
TFAP2A	c.52-1352G>A	.	intron	RP1	c.1625C>G	p.Ser542*	stop gained	0.99945
GJA1	c.*1119_*1120insA	.	3 prime UTR	RP1	c.1625C>G	p.Ser542*	stop gained	0.99940
PHC1	c.115-427G>T	.	intron	RP1	c.1625C>G	p.Ser542*	stop gained	0.99937
BEST1	c.*1721G>A; c.*1692G>A	.; .	3 prime UTR; 3 prime UTR	TRPM1	c.1879G>A	p.Val627Met	missense	0.99937
APC2	c.1208-67_1208-66insGGGGGGGGG; c.967G>C	.; p.Gly323Arg	intron; missense	RP1	c.1625C>G	p.Ser542*	stop gained	0.99925
TBR1	c.1190+182_1190+184delTTT	.	intron	RP1	c.1625C>G	p.Ser542*	stop gained	0.99923
TRPM1	c.1879G>A	p.Val627Met	missense	RBP3	c.2919C>A	p.Ser973Ser	synonymous	0.99922
RAD54B	c.945-1791_945-1789delAAA	.	intron	RP1	c.1625C>G	p.Ser542*	stop gained	0.99920
ERCC1	c.426-9C>T	.	intron	RP1	c.1625C>G	p.Ser542*	stop gained	0.99917

Table reports, for each digenic combination classified as pathogenic and for each gene included in the gene pair, the gene symbol and the HGVS of each variant referred to the specific gene (could be one heterozygous or homozygous variant, or two variants assumed in compound heterozygosity). A digenic combination is classified as pathogenic for the reported phenotypes if the associated score is greater than an optimized threshold of 0.13587435. If more than 50 digenic combinations are classified pathogenic by DIVAs, only the first 50 pathogenic ones are reported.

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