Supplemental table 3. Regional subgroup analysis of allele frequency in 24 prevalent likely pathogenic variants between 6 institutions in the USA

Nucleotide change, amino acid change/effect	JHU	GBMC	PENN	RFSW	MEC	CC	USA total
c.5882G>A, p.Gly1961Glu	16.70%	6.00%	18.80%	16.70%	4.20%	16.70%	13.64%
c.2588G>C, p.Gly863Ala	4.80%	4.00%	6.30%	10.40%	12.50%	6.70%	7.02%
c.5461-10T>C, splice site alteration	9.50%	8.00%	2.10%	0.00%	4.20%	6.70%	4.96%
c.4139C>T, p.Pro1380Leu	2.40%	10.00%	2.10%	4.20%	4.20%	0.00%	4.13%
c.1622T>C, p.Leu541Pro	0.00%	6.00%	4.20%	4.20%	0.00%	3.30%	3.31%
c.4577C>T, p.Thr1526Met	0.00%	0.00%	4.20%	4.20%	0.00%	3.30%	2.07%
c.6320G>A, p.Arg2107His	4.80%	4.00%	0.00%	2.10%	0.00%	0.00%	2.07%
c.4919G>A, p.Arg1640Gln	0.00%	0.00%	2.10%	2.10%	0.00%	6.70%	1.65%
c.4918C>T, p.Arg1640Trp	0.00%	0.00%	0.00%	2.10%	8.30%	3.30%	1.65%
c.5714+5G>A, splice site alteration	2.40%	0.00%	4.20%	0.00%	0.00%	3.30%	1.65%
c.6079C>T, p.Leu2027Phe[†]	7.10%	0.00%	0.00%	0.00%	0.00%	3.30%	1.65%
c.768G>T, p.Val256Val (splice site alteration)	0.00%	2.00%	4.20%	2.10%	0.00%	0.00%	1.65%
c.3322C>T, p.Arg1108Cys	4.80%	2.00%	2.10%	0.00%	0.00%	0.00%	1.65%
c.6089G>A, p.Arg2030Gln	0.00%	0.00%	2.10%	0.00%	0.00%	6.70%	1.24%

c.4253+4C>T, splice site alteration †	0.00%	0.00%	0.00%	0.00%	12.50%	0.00%	1.24%
c.3259G>A, p.Glu1087Lys†	2.40%	0.00%	0.00%	0.00%	8.30%	0.00%	1.24%
c.634C>T, p.Arg212Cys	0.00%	4.00%	0.00%	2.10%	0.00%	0.00%	1.24%
c.454C>T, p.Arg152Ter	2.40%	0.00%	4.20%	0.00%	0.00%	0.00%	1.24%
c.1A>G, p.Met1Val (splice site alteration)	4.80%	2.00%	0.00%	0.00%	0.00%	0.00%	1.24%
c.2966T>C, p.Val989Ala	4.80%	2.00%	0.00%	0.00%	0.00%	0.00%	1.24%
c.160+5G>A, splice site alteration†	0.00%	0.00%	0.00%	0.00%	8.30%	0.00%	0.83%
c.5395A>G, p.Asn1799Asp†	0.00%	0.00%	0.00%	4.20%	0.00%	0.00%	0.83%
c.2977_2984delGACATTGA, p.Asp993AsnfsTer27	0.00%	4.00%	0.00%	0.00%	0.00%	0.00%	0.83%
c.283T>C, p.Ser95Pro	4.80%	0.00%	0.00%	0.00%	0.00%	0.00%	0.83%

JHU = The Wilmer Eye Institute, Johns Hopkins University, Baltimore, MD; GBMC = Greater Baltimore Medical Center, Baltimore, MD; PENN = Sheie Eye Institute, University of Philadelphia, Philadelphia, PA; RFSW = Retina Foundation of the Southwest, Dallas; MEC = Moran Eye Center, Salt Lake City, UT; CC = Cole Eye Institute, Cleveland Clinic, Cleveland, OH.

†Comparison analysis revealed statistical difference in 5 variants.

121 patients from USA harboring multiple likely pathogenic variants consist of 21 from JHU, 25 from GBMC, 24 from PENN, 24 from RFSW, 12 from MEC, and 15 from CC.

The high allele frequency shown on black background was defined as the allele frequency of at least 2.0% in each subgroup.