Supplemental table 4. Subgroup analysis of allele frequency in 9 prevalent likely pathogenic variants between 152 females and 127 males

Nucleotide change, amino acid change/effect	Female	Male	Total number of detected alleles in ProgStar cohort	Allele frequency in female	Allele frequency in male
c.5882G>A, p.Gly1961Glu	47	37	84	15.46%	14.57%
c.2588G>C, p.Gly863Ala	18	22	40	5.92%	8.66%
c.5461-10T>C, splice site alteration	15	12	27	4.93%	4.72%
c.4139C>T, p.Pro1380Leu	13	9	22	4.28%	3.54%
c.1622T>C, p.Leu541Pro	6	9	15	1.97%	3.54%
c.3322C>T, p.Arg1108Cys	7	6	13	2.30%	2.36%
c.5714+5G>A, splice site alteration	10	3	13	3.29%	1.18%
c.6079C>T, p.Leu2027Phe	5	8	13	1.64%	3.15%
c.6089G>A, p.Arg2030Gln†	8	1	9	2.63%	0.39%

†Comparison analysis revealed statistical difference in 1 variant.

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