Appendix 3. Remaining variants after performing the filtering steps.

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| Patient | Gene | Accession number | Mutation Type | Nucleotide change | Predicted effect | Genotype | SNP ID |
| JU#1303 | *RPE65* | NM\_000329.2 | Nonsense | c.1028T>A | p.(L343\*) | Heterozygous | None |
|  |  | Missense | c.683A>C | p.(Q228P) | Heterozygous | rs886046510 |
| *NBAS* | NM\_015909.3 | Missense | c.3407A>T | p.(H1136L) | Heterozygous | rs183391101 |
|  |  | Missense | c.1648G>T | p.(G550C) | Heterozygous | rs571873632 |
| JU#1085 | *RPE65* | NM\_000329.2 | Missense | c.1543C>T | p.(R515W) | Heterozygous | rs121917745 |
|  |  | Missense | c.683A>C | p.(Q228P) | Heterozygous | rs886046510 |
| *ALMS1* | NM\_015120.4 | Missense | c.9925C>T | p.(P3309S) | Heterozygous | None |
|  |  | Missense | c.11556T>A | p.(H3852Q) | Heterozygous | rs186141821 |