**Appendix 3: Distribution of Synonmous coding region variants in LHON cases**

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| **S.No** | **Familly ID** | **Nucleotide substitution** | **Amino acid change** | **Codon change** | **Amino acid change** | **Novel/Reported** | **Locus change** | **Disease associated** |
|  | LH-40 | m.3447A>G | p.Gln47Gln | CAA>CAG | Gln>Gln | Reported | *ND1* | Nil |
|  | LH-70 | m.3535T>C | p.Leu77Leu | TTA>CTA | Leu>Leu | Reported | *ND1* | Nil |
|  | LH-39 | m.3591G>A | p.Leu95Leu | CTG>CTA | Leu>Leu | Reported | *ND1* | Nil |
|  | LH-57 | m.3693G>A | p.Leu129Leu | CTG>CTA | Leu>Leu | Reported | *ND1* | Nil |
|  | LH-30 | m.3720A>G | p.Gln138Gln | CAA>CAG | Gln>Gln | Reported | *ND1* | Nil |
|  | LH-28, LH-65 | m.3741C>T | p.Thr145Thr | ACC>ACT | Thr>Thr | Reported | *ND1* | Nil |
|  | LH-71 | m.3777T>C | p.Ser157Ser | AGT>AGC | Ser>Ser | Reported | *ND1* | Nil |
|  | LH-28,46 | m.3834G>A | p.Leu176Leu | CTG>CTA | Leu>Leu | Reported | *ND1* | Nil |
|  | LH-30 | m.3849G>A | p.Leu181Leu | TTG>TTA | Leu>Leu | Reported | *ND1* | Nil |
|  | LH-12 | m.3882G>A | p.Glu192Glu | GAG>GAA | Glu>Glu | Reported | *ND1* | Nil |
|  | LH-52,62,47 | m. 3921 C>T | p.Ser205Ser | TCC>TCT | Ser>Ser | Reported | *ND1* | Nil |
|  | LH-30 | m.3960C>T | p.Gly218Gly | GGC>GGT | Gly>Gly | Reported | *ND1* | Nil |
|  | LH-48 | m.11362A>G | p.Met201Met | ATA>ATG | Met>Met | Reported | *ND4* | Nil |
|  | LH-25 | m.11413A>G | p.Lys218Lys | AAA>AAG | Lys>Lys | Novel | *ND4* | Nil |
|  | LH-57 | m.11431C>T | p.Pro224Pro | CCC>CCT | Pro>Pro | Reported | *ND4* | Nil |
|  | LH-20 | m.11437T>C | p.Gly226Gly | GGG>GGA | Gly>Gly | Reported | *ND4* | Nil |
|  | LH-35,LH-43 | m.11467A>G | p.Leu236Leu | TTA>TTG | Leu>Leu | Reported | *ND4* | Altered brain Ph/sCJD patients |
|  | LH-51 | m.11485T>C | p.Gly242Gly | GGT>GGC | Gly>Gly | Reported | *ND4* | Nil |
|  | LH-52 | m.11722T>C | p.Leu321Leu | CTT>CTC | Leu>Leu | Reported | *ND4* | Nil |
|  | LH24,LH-37 | m.11735C>T | p.Leu326Leu | CTA>TTA | Leu>Leu | Novel | *ND4* | Nil |
|  | LH-61 | m.11890A>G | p.Gly377Gly | GGA>GGG | Gly>Gly | Reported | *ND4* | Nil |
|  | LH-52 | m.14179A>G | p.Tyr165Tyr | TAT>TAC | Tyr>Tyr | Reported | *ND6* | Recurrent pregnancy |
|  | LH-51 | m.14220A>G | p.Leu152Leu | TTA>CTA | Leu>Leu | Reported | *ND6* | Nil |
|  | LH-60 | m.14305G>A | p.Ser123Ser | AGC>AGT | Ser>Ser | Reported | *ND6* | Nil |
|  | LH-52 | m.14323G>A | p.Asn117Asn | AAC>AAT | Asn>Asn | Reported | *ND6* | Nil |
|  | LH-45 | m.14440A>G | p.Tyr78Tyr | TAT>TAC | Tyr>Tyr | Novel | *ND6* | Nil |
|  | LH-64 | m.14470T>C | p.Gly68Gly | GGA>GGG | Gly>Gly | Reported | *ND6* | Nil |
|  | LH-55 | m.14536A>G | p.Phe46Phe | TTT>TTC | Phe>Phe | Reported | *ND6* | Diabetes mellitus and deafness |
|  | LH-28,LH-43 | m.14569G>A | p.Ser35Ser | AGC>AGT | Ser>Ser | Reported | *ND6* | Nil |
|  | LH-15 | m.14587A>G | p.Gly29Gly | GGT>GGC | Gly>Gly | Reported | *ND6* | Nil |
|  | LH-60 | m.14590C>T | p.Gly28Gly | GGG>GGA | Gly>Gly | Reported | *ND6* | Nil |
|  | LH-70 | m.14629C>T | p.Gly15Gly | GGG>GGA | Gly>Gly | Reported | *ND6* | Nil |