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| **Table S2.** Putative pathogenic variants detected by WES in the LCA genes. | | | | | | | | |
| **Gene** | **GenBank ID** | **Position (GRCh38/hg38)** | **Nucleotide change** | **Amino acid change** | **Zygosity** | **Variant Effect** | **Existing Variation** | **MAF** |
| *ALMS1* | NM\_015120.4 | chr2:73422928 | c.718C>T | p.Pro240Ser | Hetero | missense variant | rs1320912703 | 0,000008 (GnomAD\_exome) |
| *LCA5* | NM\_181714.3 | chr6:79487263 | c.1835G>C | p.Ser612Thr | Hetero | missense variant | rs764235088 |  |
| *NMNAT1* | NM\_022787.3 | chr1:9981175 | c.439+5G>T | p.? | Hetero | splice region variant |  |  |
| Hetero: heterozygous; MAF: minor allele frequency. | | | | | | | | |