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| ***ATP6V1A* genomic DNA primers Exon 4 & Exon 13** | |
| Exon 4 LEFT PRIMER | TGCTACTCAGTTGTTAGGACGT |
| Exon 4 RIGHT PRIMER | AGTTCCTCTGTTTCGTGGGG |
| Exon 13 LEFT PRIMER | TTGCCACCATCAATCTTACA |
| Exon 13 RIGHT PRIMER | CCTTCACCAAAACACAACAGA |

Primer sequences used for validation and co-segregation analyses.

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| **cDNA sequencing of *ATP6V1A* Exon 4 – Exon 13** | |
| hseqATP6V1A Exon 4 F | CGCACTGGTAAACCCCTCTC |
| hseqATP6V1A Exon 4 R | TTGCAAGGTGTAAAGTCCCA |
| hseqATP6V1A Exon 13 F | GTTCGTTCCTCTGAGGACGA |
| hseqATP6V1A Exon 13 R | ACAGCATCCCTACTGTCTTGT |

Primer sequences used for sequencing the variants c.317T>C and c.1513\_1514del in *ATP6V1A on cDNA*

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| **qPCR primers *ATP6V1A* Exon 1-2** | |
| qhATP6V1A Ex1-2 F: | TGGTTATGTGCATGGGGTCT |
| qhATP6V1A Ex 1-2 R: | AGTAGCCATGTCACCCTCCA |

Primer sequences used for quantitative RT-PCR to determine *ATP6V1A expression*