

Table 1: Exonic variants selected from this study in *SLC4A1*, *ATP6V1B1* and *ATP6V0A4* and results of Silico Analyses.

Gene	Variant		Exon	Exon length	Bp from	BDGP		HSF	
						3'	5'	Gained	Disrupted
<i>SLC4A1</i>	c.1437C>G	p.Cys479Trp	13	195	+6	0.85	0.80	4	2
<i>SLC4A1</i>	c.1564G>A	p.Glu522Lys	13	195	-63	0.85	0.80	2	4
<i>SLC4A1</i>	c.1765C>A	p.Arg589Cys	14	174	-34	0.97	0.88	2	4
<i>ATP6V1B1</i>	c.368G>T	p.Gly123Val	5	78	+1	0.96	0.98	3	0
<i>ATP6V1B1</i>	c.370C>T	p.Arg124Trp	5	78	+3	0.96	0.98	3	0
<i>ATP6V1B1</i>	c.481G>A	p.Glu161Lys	6	140	+36	0.82	0.99	4	3
<i>ATP6V1B1</i>	c.484G>T	p.Glu162*	6	140	+38	0.82	0.99	2	5
<i>ATP6V1B1</i>	c.1102G>A	p.Glu368Lys	11	83	+43	0.89	0.92	0	7
<i>ATP6V1B1</i>	c.1251C>G	p.Tyr417*	13	130	+3	0.66	1	2	6
<i>ATP6V0A4</i>	c.52C>T	p.Gln18*	3	117	+53	NA	0.67	3	4
<i>ATP6V0A4</i>	c.322C>T	p.Gln108*	6	126	+31	0.96	NA	2	5
<i>ATP6V0A4</i>	c.1571C>T	p.Pro524Leu	15	94	-2	0.91	0.80	2	0
<i>ATP6V0A4</i>	c.1572G>A	p.Pro524Pro	15	94	-1	0.91	0.80	0	0
<i>ATP6V0A4</i>	c.2190C>G	p.Tyr730*	20	118	+51	0.42	0.94	5	4
<i>ATP6V0A4</i>	c.2257C>T	p.Glu753*	20	118	-1	0.42	0.94	3	0