

Supplementary Table 1: List of clinically significant variants of HPS6 gene

S.No	SNP ID	Clinical Significance	Variation	Type
1	rs1590262450	Pathogenic	NM_024747.5(HPS6):c.155delT (p.Val52fs)	Frameshift
2	rs1590263807	Pathogenic	NM_024747.5(HPS6):c.1624delG (p.Asp542fs)	Frameshift
3	rs1131692332	Pathogenic	NM_024747.5(HPS6):c.1898delC (p.Pro633fs)	Frameshift
4	rs1131692333	Pathogenic	NM_024747.5(HPS6):c.2038C>T (p.Gln680Ter)	Nonsense
5	rs1220869113	Pathogenic	NM_024747.5(HPS6):c.1711_1712insAG (p.Cys571Ter)	Nonsense
6	rs1564899492	Pathogenic	NM_024747.5(HPS6):c.1065dupG (p.Leu356fs)	Frameshift
7	rs281865107	Pathogenic	NM_024747.5(HPS6):c.223C>T (p.Gln75Ter)	Nonsense
8	rs281865109	Pathogenic	NM_024747.5(HPS6):c.815C>T (p.Thr272Ile)	Missense
9	rs281865110	Pathogenic	NM_024747.5(HPS6):c.913C>T (p.Gln305Ter)	Nonsense
10	rs281865112	Pathogenic	NM_024747.5(HPS6):c.1234C>T (p.Gln412Ter)	Nonsense
11	rs281865113	Pathogenic	NM_024747.5(HPS6):c.1714_1717delTGTC (p.Leu572fs)	Frameshift
12	rs281865114	Pathogenic	NM_024747.5(HPS6):c.1865_1866delTG (p.Leu622fs)	Frameshift
13	rs1564899012	Pathogenic	NM_024747.5(HPS6):c.283delG (p.Val95fs)	Frameshift
14	rs1564899951	Pathogenic	NM_024747.5(HPS6):c.1864_1871delGCTCTGGA (p.Leu622fs)	Frameshift
15	rs1478574193	Likely Pathogenic	NM_024747.5(HPS6):c.779G>A (p.Gly260Glu)	Missense
16	rs1590263820	Likely Pathogenic	NM_024747.5(HPS6):c.1649delG (p.Gly550fs)	Frameshift
17	rs756325364	Likely Pathogenic	NM_024747.6(HPS6):c.823C>T (p.Pro275Ser)	Missense
18	rs756471925	Likely Pathogenic	NM_024747.6(HPS6):c.706_707delTC (p.Ser236fs)	Frameshift
19	1050563	Likely Pathogenic	NM_024747.6(HPS6):c.206_210dupCGGGC (p.Trp71fs)	Frameshift
20	996365	Likely Pathogenic	NM_024747.6(HPS6):c.1999C>T (p.Arg667Ter)	Nonsense
21	996366	Likely Pathogenic	NM_024747.6(HPS6):c.335G>A (p.Trp112Ter)	Nonsense
22	996367	Likely Pathogenic	NM_024747.6(HPS6):c.1732C>T (p.Arg578Ter)	Nonsense
23	rs1554903728	Likely Pathogenic	NM_024747.5(HPS6):c.1693T>G (p.Phe565Val)	Missense
24	rs1590262288	Likely Pathogenic	NM_024747.6(HPS6):c.19_20delCT (p.Leu7fs)	Frameshift
25	rs763073715	Likely Pathogenic	NM_024747.6(HPS6):c.1A>G (p.Met1Val)	Missense