

Supplementary Table 1 Detailed information on genetic variants in patients with Mucopolysaccharidosis type VI

I D	SNP	Nucleotide; protein change found	rsID	Exon s of IDS gene/ 8	Type of nucleotide change	Significan ce	Previous ly reported	Reference
1	c.1601A > G	p.Ter534Trp	rs15540696 55	8	Missense	Pathogenic	Yes	doi: 10.22038/IJBMS.2018.27742. 6760
2	c.691-1G > A	-	rs77886834 8		Splicing substitutio ns	Likely Pathogenic	Yes	doi: 10.1002/humu.23613
3	c.454C > T	p.Arg152Trp	rs99110452 5	3	Missense	Pathogenic	Yes	doi: 10.3389/fmolb2021780184
4	c.966G > A	p.Trp322*	rs15540793 18	6	Nonsense	Pathogenic	Yes	doi: 10.1002/humu.23613
5	c.194C > T	p.Ser65Phe	rs12333318	2	Missense	VUS	Yes	doi:

14	c.262C > T	p.Gln88*	rs12992078 31	1	Nonsense	Pathogenic	Yes	doi: 10.1002/humu.23613
15	c.1079T > C	p.Leu360Pro	rs15540792 84	5	Missense	Pathogenic	Yes	doi: 10.1002/humu.23613
16	c.743delC	p.Pro248fs	rs43190549 4	4	Frameshift mutations	Pathogenic	Yes	doi: 10.1002/humu.23613
17	c.785dupA	p.Asn262fs	rs74901524 6	4	Frameshift mutations	Likely Pathogenic	Yes	doi: 10.1002/humu.23613
18	c.293T > C	p.Leu98Pro	rs15540320 90	1	Missense	Pathogenic	Yes	doi: 10.1002/humu.23613
19	c.1131G > T	p.Trp377Cys	-	5	Missense	VUS	No	
20	c.941T > C	p.Leu314Pro	-	5	Missense	VUS	Yes	doi: 10.3389/fmolb2021780184
21	ex5: del2635kb	-	-		Large deletion	Pathogenic	No	
22	c.990_1003del14	-	-		Small deletion	Pathogenic	No	

VUS: variant of unknown significance.