Table 2: The most common variants in patients with Myotonia Congenita (*CLCN1*) in UK

CLCN1 Variant	Patients, n (% of total)	AD	AR
p.Gly230Glu†	154 (18)	154	0
c.180+3A>T (splice site)†	90 (10)	0	84
p.Trp303Arg†	59 (7)	59	0
p.Gly285Glu†	46 (5)	1	26
p.Ala313Thr†	44 (5)	44	0
p.Arg894X†	37 (4)	4	11
p.Phe167Ser	29 (3)	0	27
p.Phe307Ser†	24 (3)	24	0
p.Phe297Ser†	23 (3)	23	0
c.1437_1450del p.Pro480fs†	22 (3)	0	20
p.Phe413Cys†	17 (2)	0	9
p.Val327Ile†	16 (2)	0	8
p.Phe306Leu†	16 (2)	16	0
p.Met485Val†	12 (1)	0	8
p.Ala313Val†	11 (1)	11	0
p.Ala566Thr	10 (1)	0	10
c.1471+1G>A ;p.?	10 (1)		**
p.Pro480His	8 (0.9)		**
p.Glu624fs	8 (0.9)	0	8
p.Pro883Thr	8 (0.9)	0	8
p.Gly190Arg	8 (0.9)	1	6
p.Gly276Asp	7 (0.8)	6	0

Abbreviations: AD=autosomal dominant; AR = autosomal recessive
[†]previously reported *CLCN1* variants in 2011 prevalence study; [**]undetermined, likely AR