

Supplementary Table: Summary of clinical and mutational data of the 32 patients diagnosed for GNE myopathy

Patient ID	Ethnic Origin	Gender	Clinical presentation	Age of onset	Muscle Biopsy Findings	Nucleotide Substitution (Variant status ^a)	GNE exon ^b	Amino-acid Substitution (GNE protein domain ^c)
						mRNA Variant 1 NM_001128227.2		hGNE2 NP_001121699.1
P1	Arabian Peninsula	F	Distal weakness of lower and upper limbs evolving to proximal weakness with quadriceps sparing	23	Not stated	c.2228T>C (HOZ)	13	p.Met743Th (kin)
P2	Middle East	F	Distal weakness of lower limbs evolving to proximal weakness	18	Not stated	c.2129T>G (HTZ)	13	p.Val710Gly (kin)
						c.2179G>A (HTZ)	13	p.Val727Met (kin)
P3	Caucasian	F	Distal weakness of lower limbs	Not stated	Not stated	c.977G>A (HTZ)	6	p.Gly326Asp (ep-AR)
						c.1218A>C (HTZ)	8	p.Lys406Asn (ep)
P4	Caucasian	F	Distal weakness of lower limbs	41	Rimmed vacuoles	c.1985C>T (HOZ)	12	p.Ala662Val (kin)
P5	Caucasian	M	Proximal and distal weakness of lower and upper limbs	18	Rimmed vacuoles	c.860C>T (HTZ)	5	p.Ala287Val (ep-AR)
						c.2179G>A (HTZ)	13	p.Val727Met (kin)
P6	North Africa	F	Distal weakness of lower limbs	Not stated	Rimmed vacuoles	c.173C>T (HOZ)	3	p.Pro58Leu (ep)
P7	Caucasian	F	Distal weakness initially of the upper limbs evolving to lower limbs with quadriceps sparing	34	Rimmed vacuoles	c.946G>C (HTZ)	6	p.Asp316His (ep-AR)
						c.1985C>T (HTZ)	12	p.Ala662Val (kin)
P8	Caucasian	M	Distal weakness of lower limbs	37	Rimmed vacuoles	c.986T>C (HTZ)	6	p.Ile329Thr (ep-AR)
						c.1618C>T (HTZ)	10	p.His540Tyr (kin)
P9	Not stated	F	Distal weakness of lower limbs	30	Rimmed vacuoles	c.2179G>A (HOZ)	13	p.Val727Met (kin)
P10	Caucasian	M	Proximal and distal weakness with quadriceps sparing	37	Not stated	c.1937 C>G (HTZ)	12	p.Ser646X (kin)
						c.1985C>T (HTZ)	12	p.Ala662Val (kin)
P11	Turkey	F	Proximal and distal weakness of lower limbs with quadriceps sparing	24	Rimmed vacuoles	c.1618C>T (HTZ)	10	p.His540Tyr (kin)
						c.1769G>C (HTZ)	11	p.Gly590Ala (kin)
P12	India	M	Distal weakness of lower limbs	18	Rimmed vacuoles	c.986T>C (HTZ)	6	p.Ile329Thr (ep-AR)
						c.1853T>C (HTZ)	11	p.Ile618Thr (kin)
P13	Caucasian	M	Distal weakness of lower limbs	Not stated	Rimmed vacuoles	c.922C>T (HTZ)	6	p.Arg308Cys (ep-AR)
						c.1054C>T (HTZ)	6	p.Arg352Cys (ep)
P14	Caucasian	M	Distal weakness of lower limbs	30	Not stated	c.1835G>T (HOZ)	11	p.Cys612Phe (kin)
P15	French Guiana	M	Distal weakness of lower limbs	36	Rimmed vacuoles	c.717T>G (HTZ)	5	p.Asp239Glu (ep)
						c.752A>C (HTZ)	5	p.His251Pro (ep)
P16	Caucasian	M	Distal weakness of lower limbs evolving to proximal weakness (lower and upper limbs)	18	Rimmed vacuoles	c.709G>A (HTZ)	4	p.Gly237Ser (ep)
						c.1183G>A (HTZ)	8	p.Gly395Arg (ep)
P17	North Africa	M	Distal weakness of lower limbs evolving to proximal weakness	35	Rimmed vacuoles	c.679G>T (HOZ)	4	p.Asp227Tyr (ep)

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						mRNA Variant 1 NM_001128227.2		hGNE2 NP_001121699.1
P18	North Africa	M	Distal weakness of lower limbs with quadriceps sparing	27	Rimmed vacuoles	c.1853T>C (HTZ)	11	p.Ile618Thr (kin)
						c.2179G>A (HTZ)	13	p.Val727Met (kin)
P19	Caucasian	F	Distal weakness of lower limbs evolving to proximal weakness	29	Rimmed vacuoles	c.537T>G (HTZ)	4	p.His179Gln (ep)
						c.922C>T (HTZ)	6	p.Arg308Cys (ep-AR)
P20	Caucasian	F	Distal weakness of lower limbs with quadriceps sparing	50	No rimmed vacuoles	c.922C>T (HTZ)	6	p.Arg308Cys (ep-AR)
						c.1850G>A (HTZ)	11	p.Cys617Tyr (kin)
P21	Mauritius island	F	Distal weakness of lower limbs with quadriceps sparing	22	Rimmed vacuoles	c.268C>T (HTZ)	4	p.Arg90X (ep)
						c.2179G>A (HTZ)	13	p.Val727Met (kin)
P22	Caucasian	M	Distal weakness of lower limbs	Not stated	Not stated	c.2228T>C (HOZ)	13	p.Met743Thr (kin)
P23	Caucasian	M	Distal weakness of lower limbs	Not stated	Rimmed vacuoles	c.1535G>C (HTZ)	10	p.Arg512Pro (kin)
						c.2129T>G (HTZ)	13	p.Val710Gly (kin)
P24	Middle East	M	Not stated	Not stated	Not stated	c.2228T>C (HOZ)	13	p.Met743Thr (kin)
P25	Caucasian	F	Distal weakness of lower limbs evolving to proximal weakness	40	No rimmed vacuoles	c.1225G>T (HTZ)	8	p.Asp409Tyr (ep)
						c.2045T>G (HTZ)	13	p.Leu682Arg (kin)
P26	Caucasian	F	Distal weakness of lower limbs evolving to proximal weakness	20	Rimmed vacuoles	c.830G>A (HTZ)	5	p.Arg277Gln (ep)
						c.1768G>A (HTZ)	11	p.Gly590Arg (kin)
P27	Middle East	M	Distal weakness of lower limbs with quadriceps sparing	20	Not stated	c.830G>A (HTZ)	5	p.Arg277Gln (ep)
						c.2129T>G (HTZ)	13	p.Val710Gly (kin)
P28	India	M	Distal weakness of lower limbs with quadriceps sparing evolving to proximal weakness	20	Rimmed vacuoles	c.971A>G (HOZ)	6	p.His324Arg (ep-AR)
P29	North Africa	F	Distal weakness of lower limbs	22	Rimmed vacuoles	c.2228T>C (HOZ)	13	p.Met743Thr (kin)
P30	West Africa	M	Distal weakness of lower limbs with quadriceps sparing	Not stated	Not stated	c.1984G>A (HOZ)	12	p.Ala662Thr (kin)
P31	Caucasian	F	Distal weakness of lower limbs	Childhood	Rimmed vacuoles	c.922C>T (HTZ)	6	p.Arg308Cys (ep-AR)
						c.1850G>A (HTZ)	11	p.Cys617Tyr (kin)
P32	Caucasian	F	Distal weakness of lower limbs with quadriceps sparing evolving to proximal weakness	33	Rimmed vacuoles	c.922C>T (HTZ)	6	p.Arg308Cys (ep-AR)
						c.1664C>T (HTZ)	10	p.Ala555Val (kin)

^a**Variant status:** HOZ: homozygous; HTZ: compound heterozygous

^b**GNE exon:** GNE exons were numbered as in Celeste et al. (2014)

^c**GNE protein domain:** ep: UDP-GlcNAc 2-epimerase domain; ep-AR: Allosteric region; kin: ManNAc kinase domain

Underlined: Considered in Celeste et al. as "(likely) non-disease causing Single Nucleotide Polymorphism (SNP)"

In Bold: novel pathogenic variants