Supplementary

Table S1 Genetic analysis of 14 Chinese patients with late-onset Pompe disease and pedigree validation

No.	Gene and RefSeq transcript	Nucleotide change					
		Allele 1	ACMG interpretation	Pedigree validation	Allele 2	ACMG interpretation	Pedigree validation
1	NM_000152.3 (GAA)	c.2161G>T (p.Glu721*)	P	Mother is heterozygous	c.2238G>C (p.Trp746Cys)	P	Father is heterozygous
2	NM_000152.3 (GAA)	c.1409A>G (p.Asn470Ser)	VUS	Father is heterozygous	c.1409A>G (p.Asn470Ser)	VUS	Mother is heterozygous
3	NM_000152.3 (GAA)	c.1935C>A (p.Asp645Glu)	P	Mother is heterozygous	c.2238G>C (p.Trp746Cys)	P	Father is heterozygous
4	NM_000152.3 (GAA)	c.784G>A (p.Glu262Lys)	LP	Mother is heterozygous	c.2297A>G (p.Tyr766Cys)	VUS	Father is heterozygous
5	NM_000152.3 (GAA)	c.1129G>A (p.Gly377Ser)	LP	Father didn't carry; mother wasn't tested	c.2154delC (p.Ala719Rfs*45)	P	Father is heterozygous; mother wasn't tested
6	NM_000152.3 (GAA)	c.1320_1322delGAT (p.Met440del)	LP	Mother is heterozygous	c.2238G>C (p.Trp746Cys)	P	Father is heterozygous
7	NM_000152.3 (GAA)	c.1409A>G (p.Asn470Ser)	VUS	Mother is heterozygous	c.2161G>T (p.Glu721*)	P	Father is heterozygous
8	NM_000152.3 (GAA)	c.1634C>T (p.Pro545Leu)	P	Father is heterozygous	c.1822C>T (p.Arg608*)	P	Mother is heterozygous
9	NM_000152.3 (GAA)	c.1634C>T (p.Pro545Leu)	P	Father is heterozygous	c.2242delG (p.Glu748Rfs*16)	P	Mother is heterozygous
10	NM_000152.3 (GAA)	c.2237G>A (p.Trp746*)	P	Father is heterozygous	c.2238G>C (p.Trp746Cys)	P	Mother is heterozygous
11	NM_000152.3 (GAA)	c.1299G>C (p.Gln433His)	LP	Father didn't carry; mother wasn't tested	c.2832delA (p.Glu945Sfs*78)	P	Father is heterozygous; mother wasn't tested
12	NM_000152.3 (GAA)	c.1299G>C (p.Gln433His)	LP	Father didn't carry; mother wasn't tested	c.2832delA (p.Glu945Sfs*78)	P	Father is heterozygous; mother wasn't tested
13	NM_000152.3 (GAA)	c.1879_1881delTCC (p.Ser627del)	VUS	N	c.2297A>G (p.Tyr766Cys)	LP	N
14	NM_000152.3 (GAA)	c.1634C>T (p.Pro545Leu)	P	Father didn't carry; mother wasn't tested	c.2426delC (p.Leu811fs*37)	P	Father is heterozygous; mother wasn't tested

ACMG, American College of Medical Genetics and Genomics; GAA, acid alpha-glucosidase; P, pathogenic; LP, likely pathogenic; VUS, uncertain significance; N, unknown.

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