***GAA* gene changes identified in CRIM-negative patients with Pompe disease**

Location	cDNA change	Amino acid change	Reference
Promoter-?	Multiple exon deletion		[1]
Exon 2	c.1A>G	p.Met1?	[1]
Exon 2	c.148_859-11del	p.Glu50HisfsX37	[2]
Exon 2	c.236_246del	p.Pro79ArgfsX12	[3]
Exon 2	c.340_341insT	p.Lys114fsX32	[4]
Exon 2	c.352C>T	p.Gln118X	[1]
Exon 2	c.525delT	p.Glu176ArgfsX45	[5]
Exon 2	c.525_526delTG	p.Asn177ProfsX11	[1]
Intron 2	c.546+2T>C	splice site	[6]
Exon 3	c.685_686insCGGC	p.Arg229fsProfsX102	[7]
Intron 3	c.692+1G>A	splice site	Novel
Exon 4	c.722_723delTT	p.Phe241CysfsX88	[8]
Exon 4	c.766_785delinsC	p.Tyr256ArgfsX6	[9]
Intron 4	c.858+2T>A	splice site	[1]
Exon 6	c.1075G>T	p.Gly359X	[1]
Exon 7	c.1128_1129delinsC	p.Trp376CysfsX16	[8]
Exon 7	c.1157dupA	p.Val387GlyfsX119	[10]
Exon 8-15	c.1195-18_2190-20del	p.Asp399ValfsX6	[11]
Exon 8	c.1209delC	p.Asn403LysfsX37	[1]
Exon 8	c.1292_1295dupTGCA	p.Gln433AlafsX74	Novel
Exon 10	c.1442G>A	p.Trp481X	[1]
Exon 10	c.1496G>A	p.Trp499X	[8]
Exon 10	c.1497G>A	p.Trp499X	[12]
Exon 10	c.1548G>A	p.Trp516X	[4]
Exon 11	c.1591dupG	p.Asp531GlyfsX7	[1]
Intron 11	c.1637-2A>G	splice site	[13]
Exon 12	c.1650dupG	p.Thr551AspfsX85	[1]
Exon 12	c.1654delC	p.Leu552SerfsX26	[1]
Exon 12	c.1657C>T	p.Gln553X	Novel
Exon 12	c.1687C>T	p.Gln563X	[14]
Intron 12	c.1754+1G>A	splice site	[1]
Intron 12	c.1754+2T>A	splice site	[1]
Exon 13	c.1802C>A	p.Ser601X	Novel
Exon 13	c.1822C>T	p.Arg608X	[8]
Exon 13	c.1826dupA	p.Tyr609X	[4]
Exon 13	c.1827delC	p.Tyr609X	[4]
Intron 13	c.1888+1G>A	splice site	[8]
Exons 16-20	c.2222_*549+214delins13	p.Asp741AlafsX28	Novel

Exon 16	c.2237G>A	p.Trp746X	[15]
Exon 16	c.2238G>A	p.Trp746X	[7]
Exon 16	c.2300delT	p.Phe767SerfsX14	[8]
Intron 16	c.2331+2T>A	splice site	[8]
Exon 17	c.2432delT	p.Leu811fsArgX37	[16]
Exon 17	c.2439dupC	p.Ile814HisfsX70	[1]
Exon 18	c. 2495_2496delCA	p.Thr832AsnfsX51	[13]
Exon 18	c.2544delC	p.Lys849ArgfsX39	Novel
Exon 18	c.2560C>T	p.Arg854X	[17]
Exon 18	c.2608C>T	p.Arg870X	[2]
Exon 19	c.2706delG	p.Lys903ArgfsX2	[1]

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Additional novel *GAA* gene changes identified in patients with Pompe disease

Location	DNA change	Protein change	*PolyPhen-2 prediction
Exon 2	c.40_47delGCCGTCTG	p.Ala14ArgfsX18	N/A
Exon 3	c.684dupG	p.Arg229ProfsX101	N/A
Exon 5	c.947A>G	p.Asn316Ser	Probably damaging
Exon 9	c.1402A>T	p.Ile468Phe	Probably damaging
Exon 10	c.1538G>A	p.Trp516X	N/A
Exon 13	c.1844G>A	p.Gly615Glu	Probably damaging
Exon 14	c.1913G>A	p.Gly638Glu	Probably damaging
Exon 14	c.2017A>T	p.Asn673Tyr	Probably damaging
Exon 16	c.2234T>C	p.Leu745Pro	Probably damaging
Exon 16	c.2294G>A	p.Gly765Asp	Probably damaging
Exon 17	c.2453T>C	p.Leu818Pro	Probably damaging
Exon 17	c.2474C>G	p.Pro825Arg	Probably damaging
Exon 18	c.2537C>A	p.Ala846Asp	Probably damaging

Compiled June 3, 2014

* http://genetics.bwh.harvard.edu/pph2/

N/A: Not applicable because mutation is not a missense mutation.

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