

Title: Mucopolipidosis III Alpha/Beta *GeneReview* Table 2

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Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

Table 2. *GNPTAB* Mutation Summary Table

Mutation	Alternate Name ¹	Exon	Reference
c.10A>C	174A>C	1	Kudo et al [2006]
c.118-2A>G		2	Bargal et al [2006]
c.310C>T		3	Paik et al [2005]
c.569A>T	733A>T	5	Kudo et al [2006]
Alu insert		5	Tappino et al [2008]
c.616del CAGA	773_776del CAGA	6	Kudo et al [2006]
c.771G>A		7	Steet et al [2005]
c.832C>T	996C>T	8	Kudo et al [2006]
c.848delA	1012delA	8	Kudo et al [2006]
c.1196C>T		10	Bargal et al [2006]
c.1220A>C		10	Tiede et al [2005a]
c.1580delC	1744delC	12	Kudo et al [2006]
c.1625insC		13	Tiede et al [2005b]
c.1738_1741 tripTATA	1902_1905 tripTATA	13	Kudo et al [2006]
c.1895C>G		13	Tiede et al [2005a]
c.2051_2055delACTCA	2215_2219delACTCA	13	Kudo et al [2006]
c.2188T>AAA	2352T>AAA	13	Kudo et al [2006]
c.2533C>T		13	Bargal et al [2006], Tiede et al [2005b]
c.2574_2575delGA		13	Paik et al [2005]
c.2659dupA		13	Tappino et al [2008]
c.2681G>A		13	Paik et al [2005]
c.2715+1G>A		13	Paik et al [2005]
c.2916insT		15	Bargal et al [2006]
c.3145insC		16	Tiede et al [2005b]
c.3173C>G		16	Paik et al [2005]
c.3231_3234dupCTAC	3395_3398dupCTAC	16	Kudo et al [2006]
c.3249+1G>A		16	Bargal et al [2006]
c.3252delA		17	Tiede et al [2005b]

Mutation	Alternate Name ¹	Exon	Reference
c.3335+1G>A		17	Tiede et al [2005b], Kudo et al [2006]
c.3335+6T>G	IVS17+6T>G	17	Bargal et al [2006], Kudo et al [2006]
c.3434+1G>A		18	Bargal et al [2006]
c.3435-1G>A	IVS19-1G>A	19	Kudo et al [2006]
c.3474_3475delTA		19	Paik et al [2005]
c.3503_3504delTC	3665_3666delTC	19	Plante et al [2008], Kudo et al [2006], Bargal et al [2006]
c.3565C>T		19	Paik et al [2005], Bargal et al [2006]
c.3566insA		19	Tiede et al [2005b]
c.3613C>T		20	Bargal et al [2006], Tiede et al [2005b], Kudo et al [2006]

1. The alternate name column has been added to clarify the nomenclature of Kudo et al 2006. The reference nucleotide sequence used by Kudo et al 2006 is GenBank AY687932 where the first nucleotide of the cDNA sequence is numbered 1. There are 164 nucleotides of untranslated sequence included in this numbering system. The mutation designations in the first column are numbered with the first nucleotide at the A of the ATG start codon.

References

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