Title: Mucolipidosis IV *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. Selected *MCOLN1* Mutations Not Discussed in Mucolipidosis IV *GeneReview*

c.163_197del, c.163_197insTCA
c.473_474delCC
c.1209-1210insT
c.1463_1464insGGCCGCAGCAG [1]
c.304C>T
c.514C>T
c.964C>T
c.317T>C
c.497G>T
c.1207C>T
c.1336G>T
c.1340T>C
c.1395C>G
c.1388G>A
c.236_237ins93 from NADH dehydrogenase 5 99-192 [2]
c.302_303delTC
c.920delT
c.1614delG
g.12426 A>T
c.235C>T

1. An 11-nucleotide insertion mutation causing a frame shift in exon 10 was discovered in two non-Jewish individuals [Bargal et al 2001, Altarescu et al 2002].
2. An American and a Canadian family was found to have a 93-bp insertion mutation in exon 2 that leads to altered splicing and null expression. The inserted segment was identical in sequence to part of the gene encoding mitochondrial NADH dehydrogenase subunit 5 [Goldin et al 2004].

References

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