Title: Isolated Methylmalonic Acidemia *GeneReview* – *MUT* alleles in diverse populations
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Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

MUT alleles repeatedly identified in diverse populations

- p.Gly717Val observed in 41% of affected African Americans and Nigerians is associated with a *mut*$^-$ phenotype.
- p.Gly623Arg and p.Gly94Val are recurrent pathogenic variants in African Americans [Adjalla et al 1998, Worgan et al 2006].
- p.Glu117Ter, c.385+5G>A, p.Arg369His, p.Leu494Ter, and p.Arg727Ter account for most of the pathogenic variants seen in Japan [Sakamoto et al 2007].
- The pathogenic missense variant p.Asn219Tyr was identified with increased frequency in persons of northern European heritage [Acquaviva et al 2001, Acquaviva et al 2005].
- p.Arg108Cys is a common pathogenic variant among Hispanics with *mut* class methylmalonic acidemia [Worgan et al 2006].
- p.Gly94Arg, p.Ala324Thr, c.1022dupA, p.Asn341fs, c.671-678dup, and p.Val227fs are seen in Hispanics [Martinez et al 2005, Worgan et al 2006].
- p.Gly427Asp and p.Gly544Ter have been only seen in Asians [Worgan et al 2006].
- Clusters of recurrent pathogenic variants in the same or adjacent nucleotides, associated in some cases with the presence of CpG dinucleotides, are found in exons 2, 3, 6, and 11. Three different pathogenic variants involve codon 108 (p.Arg108Cys, p.Arg108Gly, and p.Arg108His); codons 228, 369, and 694 were the site for two different pathogenic variants each (p.Arg228Ter and p.Arg228Gln; p.Arg369Cys and p.Arg369His; and p.Arg694Trp and p.Arg694Leu, respectively) [Jung et al 2005, Worgan et al 2006].
- A splice-site variant c.1636+3A>G recurs in the Arab-Moslem population [Berger et al 2001].
- One exon deletion has been documented [Acquaviva et al 2005].
- One instance of uniparental paternal isodisomy for 6p24 caused a syndrome of methylmalonic acidemia and transient neonatal diabetes mellitus by reduction to homozygosity.

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