Title: Proopiomelanocortin Deficiency *GeneReview* Table 3
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Note: The following information is provided by the authors and has not been reviewed by *GeneReviews* staff.

Table 3. *POMC* Pathogenic Allelic Variants Discussed in This *GeneReview*

DNA Nucleotide Change	Protein Amino Acid Change	References	Reference Sequences
c.7013G>T	p.Glu79Ter	Krude et al [1998]	NM_000939.2 NP_000930.1
c.7133delC	p.Arg83fs	Krude et al[1998]	
c.3804C>A	p.0	Krude et al [2003]	
c.6851A>T	p.Lys25Ter	Krude et al [2003]	
c.6996delC	p.Asn73fs	Krude et al [2003]	
c.7100insGG	p.Ser108fs	Krude et al [2003]	
c.6906delC	p.Pro69LeufsTer71	Farooqi et al [2006]	
c.6922InsC	p.Arg75fs	Clement et al [2008]	
c.231C>A	p.Tyr77Ter	Mendiratta et al [2011]	
c.202C>T	p.68GlnTer	Cirillo et al [2012]	

See Quick Reference for an explanation of nomenclature. *GeneReviews* follows the standard naming conventions of the Human Genome Variation Society (www.hgvs.org).

References

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