Title: Carnitine Palmitoyltransferase 1A Deficiency *GeneReview* Table 2

Authors: Bennett MJ, Santani AB

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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. Mutations in *CPT1A*

cDNA	Protein	Exon
Homozygous missense and nonsense mutations		
298C>T	Q100X	4
367C>T	R123C	4
986C>T	T314I	9
1069C>T	R357W	10
1361A>G	D454G	12
1436C>T	P479L	12
2129G>A	G710E	17
2126G>A	G709E	17
478C>T	R160X	5
1027T>G	F343V	10
1393G>T	G465W	12
1737C>A	Y579X	14
Heterozygous missense and nonsense mutations		
96T>G	Y32X	2
1079A>G	E360G	10
823G>A	A275T	8
912C>G	C304W	9
823G>A	A275T	8
1241C>T	A414V	11
1493A>G	Y498C	13
1069C>T	R357W	10
1451T>C	L484P	12
1425G>A	W475X	12
1494T>G	Y498X	13
946C>G?	R316G?	9
1339C>T	R446X	11
2156G>A	G719D	18

cDNA	Protein	Exon
Deletions and insertion		
Homozygous IVS 14+3kb	del 581-702	15
E525 Ins FLPYHELSX	Trunc 240 amino acids del 626-676	16
1876-1G>A		
2027-2028 +2del AAGT		17
948delG	R316fsX328	9
1600delC	L534fsX	13

Mutations are matched to the protein sequence (GI 32879925) in the PubMed protein bank [Brown et al 2001, Gobin et al 2002, Bennett et al 2004, Stoler et al 2004, Korman et al 2005, Tsuburaya et al 2010].

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