

Title: Trimethylaminuria *GeneReview*, Table 3

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Note: The following information is provided by the author(s) and has not been reviewed by GeneReviews staff.

Table 3. Rare *FMO3* Mutations Causative of TMAuria or Implicated in the Disorder

Mutation	Exon	Amino Acid Change
g.-2092 to 10145del	1 and 2	del exons 1-2
g.94G>A	2	E32K
g.110T>C	2	I37T
g.11145A>G	3	R51G
g.11148G>T	3	A52T
g.11166G>A	3	V58I
g.11177A>G	3	N61S
g.11185delA	3	K64KfsX2
g.11192G>T	3	M66I
g.11239T>C	3	M82T
g.15036A>G	4	N114S
g.15123T>A	4	V143E
g.15137G>T	4	G148X
g.15153C>T	4	P153L
g.15526_15527delTG	5	C197fsX
g.15531T>A	5	D198E
g.15533T>C	5	I199T
g.15539C>A	5	T201K
g.18177G>A	6	R223Q
g.18225G>C	6	R238P
g.21429G>T	7	E305X
g.21460G>T	7	E314X
g.21680G>T	7	R387L
g.21684G>A	7	W388X
g.21702delG	7	K394KfsX11
g.23580delG	8	M405IfsX
g.24486G>A	9	M434I
g.24592C>T	9	Q470X
g.24608G>A	9	G475D
g.24658C>T	9	R492W
g.24682C>T	9	R500X

K64KfsX2 has also been referred to as M66X

Mutation nomenclature follows that recommended by the Human Genome Organization (<http://www.hgvs.org/mutnomen/>)