Title: Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome *GeneReview* Table 5

Authors: Camacho J, Rioseco-Camacho N

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Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

Table 5. Selected *SLC25A15* Pathologic Allelic Variants

DNA Nucleotide Change	Protein Amino Acid Change	Reference
c.562_564delTTC	p.Phe188del [1, 2]	Camacho et al [1999]
c.535C>T	p.Arg179ter [1, 3]	Tsujino et al [2000]
c.538G>A	p.Glu180Lys [3, 4]	Camacho et al [1999]
c.569G>A	p.Gly190Asp [1, 2]	Salvi et al [2001]
c.564C>G	p.Phe188Leu	Tessa et al [2009]
c.95C>G	p.Thr32Arg [2, 4]	Camacho et al [2006]
c.79G>A	p.Gly27Arg [1, 3]	Salvi et al [2001]
c.658G>A	p.Gly220Arg [3, 4]	Al-Hassan et al [2008]
c.44C>A	p.Ala15Glu [1]	Shih & Ficicioglu [2000]
c.823C>T	p.Arg275ter [3]	Torisu et al [2006]
c.824G>A	p.Arg275Gln [3]	Salvi et al [2001]
c.110T>G	p.Met37Arg [3]	Tessa et al [2009]
c.818T>A	p.Met273Lys [2]	Fecarotta et al [2006]
c.337G>T	p.Gly113Cys [2]	Fecarotta et al [2006]
c.847C>T	p.Leu283Phe [1, 2]	Tessa et al [2009]
c.815C>T	p.Thr272Iso [2]	Tessa et al [2009]
c.212T>A	p.Leu71Gln [1, 2]	Tessa et al [2009]

1. *SLC25A15 (ORNT1)* mutations associated with neonatal onset

2. Residual function

3. No residual function

4. Targets normally to mitochondria

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