A Nonsense Mutation in the Exon 2 of the 3-Hydroxy- 3- (HL) Gene Producing Three Mature mRNAs Is the Main Aciduria in European Mediterranean Patients

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#	Article	IF	Citations
1	The association of nonsense codons with exon skipping. Mutation Research - Reviews in Mutation Research, 1998, 411, 87-117.	2.4	142
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18	Characterization of a novel HMG-CoA lyase enzyme with a dual location in endoplasmic reticulum and cytosol. Journal of Lipid Research, 2012, 53, 2046-2056.	2.0	8

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20	Analysis of aberrant splicing and nonsense-mediated decay of the stop codon mutations c.109G>T and c.504_505delCT in 7 patients with HMG-CoA lyase deficiency. Molecular Genetics and Metabolism, 2013, 108, 232-240.	0.5	7
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