

Title: Myoclonus-Dystonia *GeneReview* Table 3

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

Table 3. Published Mutations in the *SGCE* Gene in Individuals with Myoclonus-Dystonia

Exon	Mutation	Family History	Reference
2	164delG	Yes	Hedrich et al [2004]
2	H60P	No	Hedrich et al [2004]
3	276delG	No	Asmus et al [2002]
3	R97X	Yes	Zimprich et al 2001
3	R102X ¹	Yes ²	Zimprich et al [2001], Asmus et al [2002], Han et al [2003], Hedrich et al [2004]
3	233-1G>A	Yes	Asmus et al [2002]
4	484delAATT	Yes	Marechal et al [2003]
4	463+6T>C	No	Asmus et al [2002]
4	488-97del	Yes	Zimprich et al [2001]
5	565delA	Yes	Zimprich et al [2001]
5	625insG	Yes ²	Muller et al [2002], Kock et al [2004]
5	L196R	Yes	Klein et al [2002]
6	R237X	Yes	Doheny et al [2002]
6	733delAATT	Yes	Asmus et al [2002]
6	907+1G>A	Yes	Zimprich et al [2001]
7	832delAAAAAC	Yes	Han et al [2003]
7	835delACAAA	Yes	Klein et al [2002]
7	855insT	Yes	Foncke et al [2003]
7	966delT	Yes	Muller et al [2002]
7	974delC	Yes	Hjermind et al [2003]
7	Q286X	Yes	Asmus et al [2002]
7	1037+5G>A	Yes	Asmus et al [2002]
All	Interstitial del 7q21	No	DeBerardinis et al [2003]

1. This mutation has been reported in nine unrelated families.

2. The proband appeared to be a simplex case, but the mutation was found in their unaffected father due to reduced penetrance (only one of the R102X cases appeared simplex [Hedrich et al 2004]).

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