Title: Central Core Disease *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. *RYR1* Mutations Reported in CCD

Exon	Mutation
3	p.D60N, p.S71Y*
4	p.R109W*
6	p.R146C*
6	p.R163C, p.Q160G
7	p.G215E*
12	p.I403M, p.M402T*
13	p.S427L*, p.Q474H
14	p. E512K, p.Q512K, p.Y522S
17	p.R614C*
	p.A1577T*
40	p.R2163H, p.R2163C, p.V2168M
42	p.N2283H*
45	p.R2434H, p.M2434K*, p.R2435H, p.R2454H
46	p.I2453T, p.R2452W
47	p.R2508C
48	p.E2545D
57	p.R2939K*
67	p.K3367R
71	p.P3527S*
73	p.L3606P
91	p. R4214 del 3, p.R4214 del 16, p.A4329D*,
94	p.R4558Q*, p.L4568P
95	p.Y4631N, p.E4634K, p.T4637A, p.T4637I, p.G4638D, p.G4638N, p.G4638S, p.L4647 del 2, p.L4650P*, p.H4651P
96	p.T4709M*
97	p.K4724Q*
100	p.L4793P, p.Y4796C, p.L4796C, p.F4808P, p.L4814F, p.R4825C
101	p.A4846V*, p.V4849I*, p.V4849R, p.N4858D, p.F4860 del, p.R4861H, p.R4861C, p.F4863D del 7
102	p.Y4864C, p.H4887Y, p.G4897V, p.G4890R, p.G4891R, p.G4893R*, p.R4893P, p.R4893Q, p.R4893W, p.4894Q, p.I4898T, p.G4899E*, p.G4899R, p.A4906V, p.F4906 del, p.R4914G, p.R4914T, p.T4920N, p.F4921S, p.F4921T, p.V4927 del 2
103	p.A4940T, p.I4938M, p.N4939Q

Mutations associated with autosomal recessive inheritance are followed by an asterisk (*).
Mutations associated with rods and cores: p.Y4796C, p.G4638S, p.T4637A.
Mutations associated with fetal akinesia: p.R614C, p.L4650P, p.K4724Q, p.G4899E.
Mutations associated with severe phenotype with high penetrance phenotype: p.I4898T [Lynch et al 1999], p.Y4796C [Monnier et al 2000].
Mutation p.V4849I was found in one of 50 French controls [Monnier et al 2000], but was also found to be associated with MH [Sambuughin et al 2005] and CCD [Jungbluth et al 2002].

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