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

Table 3. Common Pathogenic Amino Acid Variants in *RYR1* Associated with Autosomal Dominant CCD

Amino Acid Change	Exon	Reference
p.R4861C p.R2508C	101 47	Davis et al 2003 Wu et al 2006
p.L4793P, p.R4825C p.R4861H p.R4893W, p.G4899E, p.R4914G	100 101 102	Monnier et al 2001
p.R2163C, p.R2163H, p.V2168M p.T2206M	38 40	Manning et al 1998

Davis MR, Haan E, Jungbluth H, Sewry C, North K, Muntoni F, Kuntzer T, Lamont P, Bankier A, Tomlinson P, Sanchez A, Walsh P, Nagarajan L, Oley C, Colley A, Gedeon A, Quinlivan R, Dixon J, James D, Muller CR, Laing NG (2003) Principal mutation hotspot for central core disease and related myopathies in the C-terminal transmembrane region of the *RYR1* gene. *Neuromuscul Disord* 13:151-7

Manning BM, Quane KA, Ording H, Urwyler A, Tegazzin V, Lehane M, et al (1998) Identification of novel mutations in the ryanodine-receptor gene (*RYR1*) in malignant hyperthermia: genotype-phenotype correlation. *Am J Hum Genet* 62:599-609

Monnier N, Romero NB, Lerale J, Landrieu P, Nivoche Y, Fardeau M, Lunardi J (2001) Familial and sporadic forms of central core disease are associated with mutations in the C-terminal domain of the skeletal muscle ryanodine receptor. *Hum Mol Genet* 10:2581-92

Wu S, Ibarra MC, Malicdan MC, Murayama K, Ichihara Y, Kikuchi H, Nonaka I, Noguchi S, Hayashi YK, Nishino I (2006) Central core disease is due to RYR1 mutations in more than 90% of patients. *Brain* 129:1470-80