

Title: Central Core Disease *GeneReview*; Table 4

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

**Table 4. 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

## References

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