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# RYR1 wt Allele

National Cancer Institute

## Source

National Cancer Institute. *RYR1 wt Allele*. NCI Thesaurus. Code C150312.

Human RYR1 wild-type allele is located in the vicinity of 19q13.2 and is approximately 154 kb in length. This allele, which encodes ryanodine receptor 1 protein, plays a role in skeletal muscle contraction and neuronal signaling. Mutation of the gene is associated with central core disease, King-Denborough syndrome, minicore myopathy with external ophthalmoplegia, congenital neuromuscular disease with uniform type 1 fiber and susceptibility to malignant hyperthermia type 1.