Open Peer Review on Qeios

SMN2 wt Allele

National Cancer Institute

Source

National Cancer Institute. <u>SMN2 wt Allele</u>. NCI Thesaurus. Code C127869.

Human SMN2 wild-type allele is located in the vicinity of 5q13.2 and is approximately 29 kb in length. This allele, which encodes survival motor neuron protein, is involved in the assembly of small nuclear ribonucleoproteins. Mutation of the gene is associated with sporadic adult-onset lower motor neuron disease (LMND) and a modified form of spinal muscular atrophy type 3.