PLEKHG5 wt Allele

National Cancer Institute

Source

National Cancer Institute. PLEKHG5 wt Allele. NCI Thesaurus. Code C150302.

Human PLEKHG5 wild-type allele is located in the vicinity of 1p36.31 and is approximately 54 kb in length. This allele, which encodes pleckstrin homology domain-containing family G member 5 protein, is involved in the regulation of nuclear factor kappa B and transforming protein RhoA signaling pathways. Mutation of the gene is associated with recessive intermediate Charcot-Marie-Tooth disease type C and autosomal recessive distal spinal muscular atrophy.