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RTEL1 wt Allele

National Cancer Institute

Source

National Cancer Institute. *RTEL1 wt Allele*. NCI Thesaurus. Code C134633.

Human RTEL1 wild-type allele is located in the vicinity of 20q13.33 and is approximately 38 kb in length. This allele, which encodes regulator of telomere elongation helicase 1 protein, is involved in telomere and chromosome maintenance. Mutation of the gene is associated with telomere-related pulmonary fibrosis and/or bone marrow failure 3 and dyskeratosis congenita types autosomal dominant 4 and autosomal recessive 5.