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APRT wt Allele

National Cancer Institute

Source

National Cancer Institute. *APRT wt Allele*. NCI Thesaurus. Code C50953.

Human APRT wild-type allele is located within 16q24 and is approximately 2 kb in length. This allele, which encodes adenine phosphoribosyltransferase protein, is involved in an AMP purine salvage reaction that recycles adenine into nucleic acids by formation of AMP from adenine and 5-phosphoribosyl-1-pyrophosphate. Defects in the APRT gene cause APRT deficiency, a disorder also known as 2,8-dihydroxyadenine urolithiasis.