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CYP27B1 wt Allele

National Cancer Institute

Source

National Cancer Institute. *CYP27B1 wt Allele*. NCI Thesaurus. Code C52353.

Human CYP27B1 wild-type allele is located within 12q13.1-q13.3 and is approximately 5 kb in length. This allele, which encodes 25-hydroxyvitamin D-1 alpha hydroxylase, mitochondrial protein, is involved in the biosynthesis of the active form of vitamin D3. Many allelic variants of the CYP27B1 gene encode proteins that are either inactive or exhibit reduced catalytic activity which, in turn, results in vitamin D-dependent rickets type I.