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COL6A3 wt Allele

National Cancer Institute

Source

National Cancer Institute. *COL6A3 wt Allele*. NCI Thesaurus. Code C150666.

Human COL6A3 wild-type allele is located in the vicinity of 2q37.3 and is approximately 90 kb in length. This allele, which encodes collagen alpha-3(VI) chain protein, is involved in beaded filament collagen formation. Mutation of the gene is associated with Bethlem myopathy, type 27 dystonia and Ullrich congenital muscular dystrophy.