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# COL1A2 wt Allele

National Cancer Institute

## Source

National Cancer Institute. *COL1A2 wt Allele*. NCI Thesaurus. Code C52480.

Human COL1A2 wild-type allele is located in the vicinity of 7q22.1 and is approximately 37 kb in length. This allele, which encodes collagen alpha-2 (I) chain protein, plays a role in the structural integrity of tendons, ligaments and bones. Mutations in the gene are associated with atypical Marfan syndrome, Ehlers-Danlos syndrome types and osteogenesis imperfecta types.