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COL1A1 wt Allele

National Cancer Institute

Source

National Cancer Institute. *COL1A1 wt Allele*. NCI Thesaurus. Code C52453.

Human COL1A1 wild-type allele is located in the vicinity of 17q21.33 and is approximately 18 kb in length. This allele, which encodes collagen alpha-1 (I) chain protein, is involved in both the development and maintenance of tendon, ligaments and bones. Mutations in the gene are associated with both Ehlers-Danlos syndrome and osteogenesis imperfecta types. In cases of dermatofibrosarcoma protuberans the gene is involved in a translocation t(17;22)(q22;q13) with the PDGF gene.