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ELN wt Allele

National Cancer Institute

Source

National Cancer Institute. *ELN wt Allele*. NCI Thesaurus. Code C75334.

Human ELN wild-type allele is located in the vicinity of 7q11.23 and is approximately 42 kb in length. This allele, which encodes elastin protein, plays a role in the regulation of the structure of the extracellular matrix. Mutations in the gene are associated with autosomal dominant cutis laxa and supravalvular aortic stenosis. Haploinsufficiency may be a factor in Williams-Beuren syndrome.