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ACTA2 wt Allele

National Cancer Institute

Source

National Cancer Institute. *ACTA2 wt Allele*. NCI Thesaurus. Code C103971.

Human ACTA2 wild-type allele is located in the vicinity of 10q23.3 and is approximately 56 kb in length. This allele, which encodes actin, aortic smooth muscle protein, is involved in smooth muscle cell contraction. Mutations of this gene predispose patients to a variety of diffuse and diverse vascular diseases, including premature onset coronary artery disease, premature ischemic strokes, familial aortic aneurysm thoracic type 6, Moyamoya disease type 5 and multisystemic smooth muscle dysfunction syndrome.