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LEMD3 wt Allele

National Cancer Institute

Source

National Cancer Institute. *LEMD3 wt Allele*. NCI Thesaurus. Code C147953.

Human LEMD3 wild-type allele is located in the vicinity of 12q14.3 and is approximately 79 kb in length. This allele, which encodes inner nuclear membrane protein Man1, plays a role in the downregulation of transforming growth factor beta (TGF- β) receptor signaling. Mutations in the gene have been associated with osteopoikilosis, Buschke-Ollendorff syndrome and melorheostosis.