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ASAH1 wt Allele

National Cancer Institute

Source

National Cancer Institute. *ASAH1 wt Allele*. NCI Thesaurus. Code C103900.

Human ASAH1 wild-type allele is located in the vicinity 8p22 of and is approximately 29 kb in length. This allele, which encodes acid ceramidase protein, is involved in ceramide metabolism. Mutation of the gene is associated with both Farber lipogranulomatosis and spinal muscular atrophy with progressive myoclonic epilepsy.