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EFEMP1 wt Allele

National Cancer Institute

Source

National Cancer Institute. *EFEMP1 wt Allele*. NCI Thesaurus. Code C106347.

Human EFEMP1 wild-type allele is located in the vicinity of 2p16 and is approximately 58 kb in length. This allele, which encodes EGF-containing fibulin-like extracellular matrix protein 1, plays a role in cell surface receptor binding. Aberrant expression of the gene is associated with glioma. Mutation of the gene is associated with Doyme honeycomb retinal dystrophy.