

Open Peer Review on Qeios

PLEC wt Allele

National Cancer Institute

Source

National Cancer Institute. <u>PLEC wt Allele</u>. NCI Thesaurus. Code C122742.

Human PLEC wild-type allele is located in the vicinity of 8q24 and is approximately 62 kb in length. This allele, which encodes plectin protein, is involved in anchoring intermediate filaments to desmosomes or hemidesmosomes. Mutations in this gene are associated with epidermolysis bullosa simplex, ogna type and epidermolysis bullosa simplex with pyloric atresia.

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