Open Peer Review on Qeios

PAFAH1B1 wt Allele

National Cancer Institute

Source

National Cancer Institute. <u>PAFAH1B1 wt Allele</u>. NCI Thesaurus. Code C75886.

Human PAFAH1B1 wild-type allele is located in the vicinity of 17p13.3 and is approximately 92 kb in length. This allele, which encodes platelet-activating factor acetylhydrolase, isoform Ib, alpha subunit protein, plays a role in the regulation of neuronal migration. Mutations in the gene are associated with lissencephaly type 1, subcortical band heterotopia, and Miller-Dieker lissencephaly syndrome.