

Open Peer Review on Qeios

CSTB wt Allele

National Cancer Institute

Source

National Cancer Institute. <u>CSTB wt Allele</u>. NCI Thesaurus. Code C128871.

Human CSTB wild-type allele is located in the vicinity of 21q22.3 and is approximately 4 kb in length. This allele, which encodes cystatin-B protein, plays a role in thiol protease inhibition. Mutation of the gene is associated with progressive myoclonic epilepsy 1A (myoclonic epilepsy of Unverricht and Lundborg).

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