

Open Peer Review on Qeios

CHAT wt Allele

National Cancer Institute

Source

National Cancer Institute. CHAT wt Allele. NCI Thesaurus. Code C104886.

Human CHAT wild-type allele is located in the vicinity of 10q11.2 and is approximately 85 kb in length. This allele, which encodes choline O-acetyltransferase protein, plays a role in the synthesis of acetylcholine. Mutation of the gene is associated with congenital myasthenic syndrome with episodic apnea (familial infantile myasthenia gravis 2).

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