Open Peer Review on Qeios

OTC wt Allele

National Cancer Institute

Source

National Cancer Institute. <u>OTC wt Allele</u>. NCI Thesaurus. Code C75883.

Human OT C wild-type allele is located in the vicinity of Xp21.1 and is approximately 69 kb in length. This allele, which encodes ornithine carbamoyltransferase, mitochondrial protein, plays a role in the mediation of the ornithine cycle. Missense, nonsense, and frameshift mutations in the gene are associated with ornithine carbamoyltransferase deficiency.