Open Peer Review on Qeios

SOX10 wt Allele

National Cancer Institute

Source

National Cancer Institute. <u>SOX10 wt Allele</u>. NCI Thesaurus. Code C102892.

Human SOX10 wild-type allele is located in the vicinity of 22q13.1 and is approximately 17 kb in length. This allele, which encodes transcription factor SOX-10 protein, plays a role in transcriptional activation. Mutation of the gene is associated with Waardenburg syndrome types 2E and 4C and with peripheral demyelinating neuropathy, central dysmyelinating leukodystrophy, Waardenburg syndrome, and Hirschsprung disease.