

Open Peer Review on Qeios

DKC1 wt Allele

National Cancer Institute

Source

National Cancer Institute. <u>DKC1 wt Allele</u>. NCI Thesaurus. Code C113620.

Human DKC1 wild-type allele is located in the vicinity of Xq28 and is approximately 15 kb in length. This allele, which encodes H/ACA ribonucleoprotein complex subunit 4 protein, plays a role in the stabilization and maintenance of telomerase and H/ACA small nucleolar RNA ribonucleoprotein biogenesis. Mutation of the gene is associated with both Hoyeraal-Hreidarsson syndrome and X-linked dyskeratosis congenita.

Qeios ID: 7LS9XU · https://doi.org/10.32388/7LS9XU