Open Peer Review on Qeios

FBN2 wt Allele

National Cancer Institute

Source

National Cancer Institute. FBN2 wt Allele. NCI Thesaurus. Code C120567.

Human FBN2 wild-type allele is located within 5q23-q31 and is approximately 401 kb in length. This allele, which encodes fibrillin-2 protein, plays a role in the formation of extracellular microfibrils and elastic fibers. Mutation of the gene is associated with both congenital contractural arachnodactyly and early-onset macular degeneration.