Open Peer Review on Qeios

PCSK9 wt Allele

National Cancer Institute

Source

National Cancer Institute. <u>PCSK9 wt Allele</u>. NCI Thesaurus. Code C122916.

Human PCSK9 wild-type allele is located in the vicinity of 1p32.3 and is approximately 25 kb in length. This allele, which encodes proprotein convertase subtilisin/kexin type 9 protein, plays a role in both receptor protein degradation and lipid metabolism. Mutation of the gene is associated with autosomal dominant familial hypercholesterolemia type 3.