## Open Peer Review on Qeios

## SMPD1 wt Allele

National Cancer Institute

## Source

National Cancer Institute. <u>SMPD1 wt Allele</u>. NCI Thesaurus. Code C105020.

Human SMPD1 wild-type allele is located within 11p15.4-p15.1 and is approximately 5 kb in length. This allele, which encodes sphingomyelin phosphodiesterase protein, is involved in ceramide biosynthesis. Mutation of the gene is associated with Niemann-Pick disease type A and type B.