

Open Peer Review on Qeios

POLE wt Allele

National Cancer Institute

Source

National Cancer Institute. <u>POLE wt Allele</u>. NCI Thesaurus. Code C106233.

Human POLE wild-type allele is located in the vicinity of 12q24.3 and is approximately 213 kb in length. This allele, which encodes DNA polymerase epsilon catalytic subunit A protein, plays a role in both DNA base-excision repair and DNA replication. Mutation of the gene may be associated with both increased susceptibility to colorectal malignancies and facial dysmorphism, immunodeficiency, livedo, and short stature (FILS) syndrome.

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