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POLE Exonuclease Domain Mutation

National Cancer Institute

Source

National Cancer Institute. *POLE Exonuclease Domain Mutation*. NCI Thesaurus. Code C131329.

A missense mutation in the POLE gene located in the region that encodes the exonuclease domain of the DNA polymerase epsilon catalytic subunit A protein (POLE protein). These mutations lead to defective DNA proof-reading by POLE protein during DNA replication and have been reported in patients with colorectal carcinoma and certain subtypes of endometrial carcinoma.