Phenotype of the NTHL1 gene mutations.

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NTHL1 gene consist of 6 exons spanning 8 kb, encodes 1067 bps length transcript. The 312 residues protein encoded by this gene is a DNA N-glycosylase of the endonuclease III family. Like a similar protein in E. coli, the encoded protein has DNA glycosylase activity on DNA substrates containing oxidized pyrimidine residues and has apurinic/apyrimidinic lyase activity. Its function is to initiate DNA base excision repair of oxidized ring saturated pyrimidine residues. Mutations of NTHL1 gene are associated with familial adenomatous polyposis 3; FAP 3 The c.268C>T (p.Gln90*) is the most common mutation described so far. Biallelic germline mutations affecting NTHL1 predispose carriers to adenomatous polyposis and colorectal cancer, but the complete phenotype is unknown. Here we present the cases of NTHL1 gene mutations curries identified in Polish polyposis registry. The most common mutation of NTHL1 gene in Polish polyposis patient was c.268C>T.