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.