Summarization of results of molecular diagnostics of colorectal polyposis in Poland.

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The polyp is any overgrowth of tissue from the surface of mucous membranes. The polyps that arise as a result of proliferative dysplasia are termed as adenomatous polyps or adenomas. They are true neoplastic lesions and are precursors of carcinoma. The hamartomatous polyps are formed as a result of abnormal mucosal maturation. They are non-neoplastic and do not have malignant potential. Hereditary diseases that produce large numbers of intestinal polyps include: familial adenomatous polyposis of the colon (MIM 175100), familial adenomatous polyposis type 2(MIM 608456), Peutz-Jeghers syndrome (MIM 175200), Juvenile polyposis syndrome (MIM 174900), and PTEN Hamartoma Tumor Syndromes (PHTS). During almost 20 years of research on polyposis in Poland we examined almost 700 families with familial adenomatous polyposis, over 30 families with Juvenile polyposis syndrome and PHTS, as well as over 40 families with Peutz-Jeghers syndrome. In our studies the mutations were detected in over 50% of families. The mutations in the genes determining the occurrence of polyposis in the Polish population are mostly heterogeneous. A close correlation between mutation type and phenotype has not been observed, however phenotypic data can be helpful in molecular diagnostics optimization.