HEREDITARY CANCER IN CLINICAL PRACTICE

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Mutations spectrum in hereditary disorders predisposing to occurrence of intestine polyposis in Poland

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The term polyp refers to any overgrowth of tissue from the surface of mucous membranes. Intestinal polyps grow out of the lining of the small and large bowels. 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. There are several hereditary diseases that produce large numbers of intestinal polyps. These disorders include: familial adenomatous polyposis of the colon (MIM 175100), familial adenomatous polyposis type 2(MIM 608456), Lynch's syndrome (MIM 120435), Peutz-Jeghers syndrome (MIM 175200), Juvenile polyposis syndrome (MIM 174900) PTEN Hamartoma Tumor Syndrome (PHTS) PHTS Includes: Bannayan-Riley-Ruvalcaba Syndrome (MIM 153480), Cowden Syndrome (MIM 153480), PTEN-Related Proteus Syndrome, Proteus-Like Syndrome. Here we present spectrum of mutation detected in over six hundred Polish families with intestinal polyposis. The studies have encompassed over 30 families with Juvenile polyposis syndrome and PHTS, over 40 families with Peutz-Jeghers syndrome and almost 600 families with familial adenomatous polyposis of the colon. The study

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