# Mutations of genes predisposing to occurrence of intestinal polyposis in Poland

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The term polyp refers to any tissue hypertrophy from the surface of mucous membranes. Intestinal polyps arise from the mucous membrane of the small and large intestines. The hyperplastic, adenomatous, hamartomatous and inflammatory polyps are main types of polyps observed in gastrointestinal tract. The adenomatous and hamartomatous polyps may occur as symptoms of susceptibility syndromes to the occurrence of neoplastic diseases. The syndromes of inherited predispositions associated with the presence of multiple intestinal polyps include: familial adenomatous polyposis (MIM 175100), Peutz-Jeghers syndrome (MIM 175200), juvenile polyposis syndrome (MIM 174900) and Cowden Syndrome(MIM 153480). The occurrence of these diseases is associated with mutations of the following genes: APC, MUTYH, STK11, BMPR1A, SMAD4 and PTEN. The spectrum of point mutations and copy number variation in genes predisposing to intestinal polyposis in the Polish population were determined. Determination of the spectrum of mutations of predisposition genes in the Polish population allows to optimize mutation detection for the Polish population. The research in part was financed by the project NCN 2013/09 / N / NZ5 / 02505