

Familial adenomatous polyposis (FAP) is a rare inherited cancer predisposition syndrome characterized by hundreds to thousands of precancerous colorectal polyps (adenomatous polyps). If left untreated, affected individuals inevitably develop cancer of the colon and/or rectum at a relatively young age. FAP is inherited in an autosomal dominant manner and caused by abnormalities (mutations) in the APC gene. Mutations in the APC gene cause a group of polyposis conditions that have overlapping features: familial adenomatous polyposis, Gardner syndrome, Turcot syndrome and attenuated FAP. Familial adenomatous polyposis affects males and females in equal numbers. It occurs in approximately one in 5,000 to 10,000 individuals in the United States and accounts for about 0.5% of all cases of colorectal cancer. One estimate suggests that familial adenomatous polyposis affects 50,000 American families. According to national registries, familial adenomatous polyposis occurs in 2.29-3.2 per 100,000 individuals. Classical FAP is diagnosed clinically when an individual has 100 or more adenomatous colorectal polyps (typically occurring by the third decade of life) or fewer than 100 polyps and a relative with FAP. Genetic testing for mutations in the APC gene is available to confirm the diagnosis of FAP and the associated conditions. Younger individuals may have fewer polyps. A diagnosis is made in younger people by the presence of the typical polyps and in immediate relative with FAP or by genetic testing.