Familial adenomatous polyposis (FAP) is an inherited disorder that typically presents with multiple polyps in the colon. These polyps become cancerous if not monitored in the early stages or if left untreated. For those with a family history of FAP, colonoscopy screenings begin in adolescence and continue throughout adulthood because polyps can arise at an early age. Colon cancer in untreated individuals develops before age 40 years. This topic warrants discussion to aid in the prompt recognition of symptom presentation. This article will address the development of FAP, its presentation, and potential treatment strategies that may be used by nurse practitioners.

AT A GLANCE

- FAP is an inherited condition typified by colorectal cancer.
- Early screening and detection is imperative for prevention of colorectal cancer in individuals and families with FAP.
- Treatment with surgery and chemotherapy is required when colorectal cancer caused by FAP is detected.

KEYWORDS
familial adenomatous polyposis; colorectal cancer; hereditary

DIGITAL OBJECT IDENTIFIER
10.1188/19.CJON.135-138

Familial Adenomatous Polyposis

Development, presentation, and treatment strategies

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Familial adenomatous polyposis (FAP), a rare hereditary condition affecting 3–10 of every 100,000 individuals worldwide, has a high malignant potential for the development of colorectal cancer and is a major cause of death for those affected (Wang et al., 2018) (see Figure 1). In addition to the development of colon and gastrointestinal tract adenomatous polyps, extracolonic cancers that may result include duodenal or gastric carcinoma, endometrial or ovarian cancer, follicular or papillary thyroid cancer, hepatoblastoma, and medulloblastoma (Lung, Trainer, Campbell, & Lipton, 2015; Perry, 2018). All three types of FAP (classic, attenuated, and autosomal recessive) are autosomal-inherited disorders that require frequent surveillance.

Classic FAP is characterized by the development of 100 or more adenomatous colorectal polyps (Lung et al., 2015). This type of FAP presents by adolescence with benign polyps numbering in the hundreds to the thousands (Perry, 2018). Over time, polyps increase in size and number, and they transform into cancer. Symptoms always present before an individual reaches age 40 years and tend to be associated with cancer development (Wang et al., 2018).

Attenuated FAP is a less aggressive variant that is characterized by fewer polyps, delayed polyp growth, and presentation at a later age (Plawski et al., 2013). In addition, attenuated FAP carries a lower risk for cancer; if this type of FAP is left untreated, colorectal cancer usually develops by the fifth or sixth decade of life (Lung et al., 2015).

Individuals with autosomal recessive FAP (AR-FAP) present with fewer than 100 polyps; these are predominately located in the large intestine (Plawski et al., 2013). Although AR-FAP is typically described as a milder form of FAP (U.S. National Library of Medicine, 2019), intestinal cancer usually presents by the sixth decade of life (Plawski et al., 2013).

Genetic Mutations

Two specific inherited genetic mutations are associated with the development of FAP and dictate the type of FAP: adenomatous polyposis coli (*APC*) and *MUTYH* (Lung et al., 2015). The most common genetic mutation involves *APC*, a tumor suppressor gene (Lung et al., 2015). Specifically, most *APC* mutations occur on chromosome 5 (Plawski et al., 2013). *APC* mutations cause unregulated cell growth and disrupt some cellular functions (e.g., cellular adhesion, cytoskeleton stabilization, apoptosis). Over time, unregulated growth promotes polyp development and transformation into cancer. *APC* mutations are observed in classic and attenuated FAP (Lung et al., 2015). The second type of mutation involves the *MUTYH* gene, which affects the ability of cells to repair oxidative DNA damage (Lung et al., 2015). The resulting cellular errors promote cell overgrowth