

People with multiple polyps may have a germline mutation that places them at higher risk for developing colorectal, gastrointestinal, and other cancers. Genetic testing can often identify the specific polyposis syndrome and provide insight into appropriate recommendations for cancer prevention and early detection. Individuals with hereditary polyposis syndromes often begin developing polyps in their teenage years and require aggressive gastrointestinal surveillance to remove polyps. For some, the polyp burden will be too high to manage endoscopically and will require risk-reducing colectomies. Identification of individuals with hereditary polyposis syndromes may help to reduce morbidity and mortality.

#### AT A GLANCE

- The identification of families with polyposis syndromes is important because aggressive surveillance and risk-reducing surgery can lead to the early detection and prevention of cancer.
- Identifying individuals at risk, referring them for genetic evaluation, and ensuring follow-up with prevention and early detection recommendations are responsibilities of oncology nurses.
- Oncology nurses can provide support and education to patients and families to help them medically and psychosocially manage their diagnoses.

#### KEYWORDS

colon polyps; polyposis syndromes; hereditary; cancer; gene mutation

#### DIGITAL OBJECT

#### IDENTIFIER

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# Hereditary Polyposis Syndromes

Opportunities for early detection in individuals and families

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Colon polyps can be a clinical component of a variety of inherited polyposis syndromes. Evaluation begins by obtaining an accurate polyp history (histologic type, number, location, and age of onset), cancer history (location, type, and age of onset), and assessment for the presence of other features (desmoid tumors, congenital hypertrophy of the retinal pigment epithelium, and thyroid tumors). Referral to a credentialed genetics professional depends on the number and histology of polyps, as well as the presence of other clinical features (Basso, Bianchi, Malesci, & Laghi, 2017). The identification of families with polyposis syndromes is important because the appropriate use of aggressive surveillance and risk-reducing surgery can lead to the early detection and prevention of colorectal and other cancers (Valle, 2017). A summary of the clinical characteristics, cancer risks, screening guidelines, and indications for genetic evaluation for common polyposis syndromes is shown in Figure 1.

## Adenomatous Polyposis Syndromes

Adenomatous polyposis often occurs because of mutations in the *APC* or *MUTYH* genes. Familial adenomatous polyposis (FAP) is characterized by cancer of the colon and rectum that is attributable to mutations in the *APC* gene. Testing for

mutations in the *APC* gene has been available since the late 1980s (Basso et al., 2017). Individuals with classic FAP may begin to develop colorectal polyps in their teenage years, often resulting in hundreds to thousands of polyps. Some people have a variant called attenuated FAP (aFAP), in which polyp growth is delayed and not as numerous (Basso et al., 2017).

Individuals with FAP may have desmoid tumors, which are fibrous tumors usually found in the tissue covering the intestines and tend to recur after surgical resection (National Comprehensive Cancer Network [NCCN], 2017). Individuals who have colon polyps and tumors outside the colon (osteomas, dental abnormalities, epidermoid cysts, fibromas, lipomas) have a variant known as Gardner syndrome (NCCN, 2017).

Although most individuals with pathogenic *APC* mutations will develop colorectal cancer, the number of polyps and the time frame depend on the specific mutation (NCCN, 2017). Identification of individuals with FAP is important because annual colonoscopy screening for this population often begins at age 10 years; consequently, children should be tested for FAP at this age (Achatz et al., 2017). Screening continues until the polyp burden becomes too high. Total proctocolectomy with ileal pouch-anal anastomosis is the preferred risk-reducing surgery (Jasperson & Burt, 2015). As many as 20% of individuals with a mutation in