Colonic polyps and polyposis syndromes in pediatric patients

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Purpose of review
Gastrointestinal polyps are commonly encountered during childhood and are one of the most common causes of rectal bleeding in this age group. Most polyps are benign and located in the colon, with the most frequent type being juvenile polyps. However, in older pediatric patients, if multiple polyps are present, in patients who have a positive family history, or if polyps are located outside of the colon, either adenomatous polyps or polyps associated with genetic abnormalities are more common.

Recent findings
Imaging techniques such as ultrasound and computed tomographic colonoscopy have recently been utilized to identify simple juvenile colonic polyps in children with rectal bleeding in whom there is a high index of suspicion. Colonoscopy with polypectomy is still required for histologic evaluation and resection of the polyp. There have been significant advances in genetic testing and management of hereditary gastrointestinal cancer syndromes with onset in childhood or adolescence that may ultimately reduce long-term morbidity and mortality. In addition to enhanced gastrointestinal and extraintestinal malignancy screening for affected individuals, specific gene mutations within a given condition such as adenomatous polyposis coli may predict clinical course and timing of specific interventions such as colectomy. In other conditions such as phosphatase and tensin homolog hamartoma tumor syndrome, phenotype may not be predicted by genotype.

Summary
Pediatricians, pediatric gastroenterologists, and adult gastroenterologists caring for children should understand how to differentiate benign polyps in the pediatric age group from those associated with a higher risk of complications including recurrence risk and risk of development of intestinal or extraintestinal malignancy. Recent advances in genetic testing, as well as development of consensus guidelines, are key in the identification, screening, and follow-up of children and adolescents with polyposis syndromes.

Keywords
familial adenomatous polyposis, juvenile polyposis syndrome, pediatric, polyp

INTRODUCTION

Gastrointestinal polyps commonly present during early childhood between 2 and 5 years of age, usually manifesting with painless rectal bleeding. Most polyps are benign, confined to the colon, and juvenile polyps by histology. In older pediatric patients especially those age 10 and older, simple juvenile polyps are less likely to be encountered and either adenomatous polyps or polyps occurring as part of a polyposis syndrome become more likely. This is especially true if polyps are present in larger numbers, are located outside of the colon, or if there is a positive family history of polyps or other conditions such as early-onset colon cancer. In this review, we will discuss the different types of polyps and associated polyposis syndromes.

CLINICAL FEATURES AND INITIAL EVALUATION

Colon polyps in children typically present with painless rectal bleeding. In some cases, there may be abdominal pain because of intussusception of a polyp or a large polyp causing obstruction, or prolapse of a pedunculated polyp (polyp on a stalk)

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KEY POINTS

- Juvenile polyps are the most frequent type of colonic polyps in the pediatric age group.
- Pediatric patients with adenomatous polyps, polyps occurring in adolescence, or multiple polyps of any type should be evaluated for a polyposis syndrome.
- There have been significant recent advances in understanding the genetic basis of the hereditary polyposis syndromes, which when fully characterized have the potential to reduce the morbidity and mortality from both gastrointestinal and extraintestinal complications and malignancies.

from the rectum. Patients with a significant polyp burden may present with iron-deficiency anemia, protein-losing enteropathy and consequent hypoalbuminemia, and diarrhea. Some children are asymptomatic, and the polyp(s) may be discovered as part of a screening for a hereditary polyposis syndrome, or as part of the workup for gastrointestinal complaints. Occasionally, polyps are discovered incidentally on imaging such as ultrasound or computed tomography performed for other indications, such as urologic evaluation [1,2].

Rectal examination may on occasion reveal a palpable, soft, and often mobile mass if the polyp is located in the rectum; in some cases, parents may observe or photograph prolapse of the polyp. Extraintestinal manifestations identifiable on physical examination of patients with the various hereditary polyposis syndromes include some combination of mucosal pigmentation of the lips, buccal mucosa, skin around the eyes, nostrils, hands and feet, and perianal region in patients with Peutz–Jeghers syndrome (PJS), subcutaneous cysts, desmoid tumors, dental abnormalities, exostosis, and pigmented ocular lesions in patients with familial adenomatous polyposis (FAP) and abnormal facies, macrocephaly, cleft lip or palate, mental retardation, or edema in patients with juvenile polyposis syndrome (JPS).

A complete blood count and iron studies may reveal an iron-deficiency anemia and a complete metabolic panel may demonstrate hypoalbuminemia especially in patients with a high polyp burden.

Colonoscopy is the preferred method of establishing the diagnosis of juvenile polyps as well as serving as the primary method to remove polyps. Polypectomy should be performed at the time of colonoscopy if well tolerated and feasible. All retrieved polyps should be sent for histologic evaluation. Recently, Wei *et al.* [3] reported a large series from China utilizing ultrasound in the evaluation of suspected polyps in childhood. Although ultrasound is diagnostic only and colonoscopy with polypectomy is still required to remove the polyp and allow for histologic evaluation, the authors reported an extremely high sensitivity and specificity rate in the detection of at least one polyp compared with the gold standard of colonoscopy [3].

In another recent series from India, intravenous contrast-enhanced computed tomography colonoscopy (IVCTC) was performed in 30 children with rectal bleeding thought to be due to a colonic polyp [4▪]. IVCTC still required a bowel preparation in all cases as well as intravenous sedation in 17% of cases [4▪]. IVCTC had a very good sensitivity but a poor specificity compared with colonoscopy in that series but the study was limited because of a poor complete colonoscopy rate. Similar to the limitations of ultrasound, in patients undergoing IVCTC, therapeutic colonoscopy is still required for polyp removal and histologic examination.

If a polyp is suspected prior to or identified at colonoscopy, a careful family history should be taken about colonic and small bowel cancer, and extraintestinal cancers. Genetic counseling and evaluation may help delineate the diagnosis of a polyposis syndrome that is discussed further below.

After removal at colonoscopy, histopathologic evaluation should be performed on all retrieved polyps. In children, gastrointestinal polyps generally fall into two major categories: adenomas and hamartomas. Solitary polyps and the majority of polyps found in the pediatric age group are usually hamartomas, which are predominantly of the juvenile type and likely benign. Solitary adenomas, on the other hand, are rare and may have long-term malignant potential and may be the initial manifestation of hereditary nonpolyposis colorectal cancer (HNPCC) also known as Lynch syndrome, which is discussed later.

In terms of hereditary polyposis syndromes occurring in the pediatric age group, FAP characterized by multiple small adenomatous polyps is more common than PJS or JPS, both of which are characterized by hamartomatous polyps. Although the polyps in both of these later two syndromes are hamartomatous, both syndromes are associated with an increased lifetime malignancy risk, which is not the case for pediatric patients with simple juvenile polyps (less than five hamartomatous polyps in lifetime without extraintestinal manifestations or a positive family history).

SIMPLE JUVENILE POLYPS

Juvenile polyps of the colon present with painless rectal bleeding or perianal polyp protrusion, on average, around 4 years of age. These polyps have

near negligible risk for malignant change and are thought to be benign. They may be single or multiple (but less than five in order to be classified as simple), and are the most frequent type found in children. The majority is located in the rectosigmoid colon, with the remainder distributed throughout the colon; a full colonoscopy is needed for a complete evaluation. Synchronous polyps are identified at colonoscopy in up to a third of cases [5,6]. In the large recent series of 487 pediatric patients by Wei *et al.* [3], 84% of the juvenile polyps were in the rectosigmoid colon, another 11% in the remainder of the left colon, 2% in the transverse colon, and 3% in the right colon. Other series have a higher frequency of transverse colon or right-sided polyps [5]. At colonoscopy, the polyps appear spherical to slightly lobular in form, with most being pedunculated with long stalks (Fig. 1). Microscopically, juvenile polyps have a Swiss cheese appearance with dilated cysts filled with mucin. At the time of colonoscopy, polyps should be removed, to alleviate symptoms and allow for histological diagnosis.

Management of juvenile polyps is determined by the number of polyps (less than five or at least five) and the absence or presence of a family history of any polyposis syndromes to distinguish between a simple juvenile polyp versus a polyposis syndrome. If, after a full colonoscopy, less than five polyps are found and there is no relevant family history, endoscopic polypectomy is considered to be sufficient treatment. Parents should be aware, however, that juvenile polyps may be the first feature of JPS, and that if new symptoms arise, the patient should undergo additional evaluation including repeat colonoscopy if indicated. If five or more juvenile polyps develop over the course of a patient's lifetime, they likely have a polyposis syndrome with associated risks rather than simple juvenile polyps.

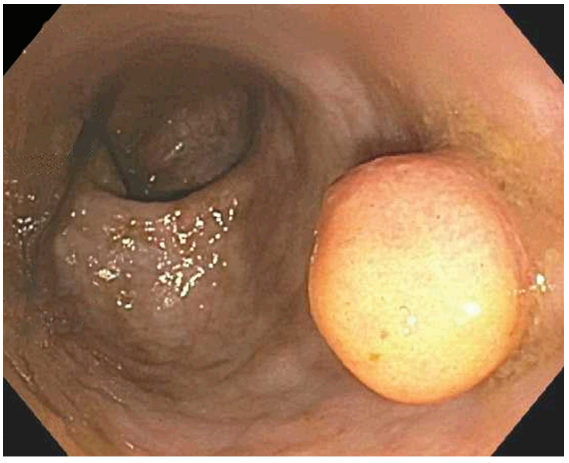

FIGURE 1. Large juvenile polyp on a stalk. The head of the polyps are typically vascular, which is why these polyps frequently present with rectal bleeding.

INFLAMMATORY POLYPS

Inflammatory polyps are commonly seen in pediatric patients with inflammatory bowel disease and other conditions associated with significant inflammation. In the majority of cases, these are actually pseudopolyps characterized by admixtures of hyperplastic and inflamed mucosa at areas of inflammation, mucosal injury, surgical anastomoses, or stricture [7▪▪]. Although they can be large, they are not premalignant and do not require removal. Inflammatory cloacogenic polyp occurs in the anorectal transition zone and appears related to solitary rectal ulcer syndrome and may present with hematochezia and tenesmus [7▪▪].

Recently, cap polyposis has been reported in children [8▪]. Macroscopically, the appearance is of small red sessile polyps covered by a fibrinopurulent exudate predominantly found at the apices of mucosal folds in the rectum or rectosigmoid; the polyps may be single or multiple and present with rectal bleeding and frequently constipation [8▪]. Histologically, the polyps are characterized by surface ulceration with a cap of fibrin inflammatory exudate. Crypts are elongated and the surrounding mucosa is macroscopically and histologically normal. Patients may resolve their symptoms following polypectomy and initiation of a stool softener, but may have a recurrence, especially in patients with a higher number of polyps [8▪]. This condition similar to solitary rectal ulcer syndrome is likely part of the mucosal prolapse spectrum of the disease.

SPECIFIC POLYPOSIS SYNDROMES IN PEDIATRIC PATIENTS

Polyposis syndromes can be divided into hamartomatous and adenomatous polyposis syndromes. The hamartomatous polyposis syndromes include JPS, phosphatase and tensin homolog (PTEN) hamartoma tumor syndrome, which includes Cowden syndrome and Bannayan–Riley–Ruvalcaba syndrome (BRRS), and PJS. These syndromes are rare with autosomal dominant transmission and variable penetrance.

Juvenile polyposis syndrome

JPS is rare with an incidence of one in 100 000–160 000 and is characterized by multiple hamartomatous polyps and an increased lifetime risk of

malignancy [9,10]. Most polyps are in the colon, although they can occur throughout the gastrointestinal tract, more commonly in the stomach than in the small bowel. The typical presentation is in childhood or adolescence, although presentation in adulthood has been reported, occasionally at the time of cancer diagnosis [9]. This diagnosis should be strongly considered in patients who have a lifetime total of five or more juvenile polyps in the colon, in patients with juvenile polyps in any other part of the gastrointestinal tract, and in patients with a juvenile polyp and a relevant family history. JPS is subclassified into three types: juvenile polyposis of infancy (which can present with diarrhea, rectal prolapse, and protein-losing enteropathy), juvenile polyposis coli (JPC) (polyps only in the colon), and generalized juvenile polyposis (polyps can involve the stomach, small bowel, and colon). Those patients with JPC and generalized juvenile polyposis can develop 50–200 polyps in their lifetime.

The typical presentation of JPC, the most common subtype, is similar to that of isolated juvenile polyps with isolated rectal bleeding; patients can be asymptomatic. The more common extraintestinal manifestations of JPS are described earlier. Other associated extraintestinal manifestations include heart defects including clinically significant mitral valve prolapse, double renal pelvis and ureter, as well as a bifid uterus and vagina. Macroscopically and histologically, the polyps are identical in appearance to a solitary juvenile polyp; polyps can also be multilobulated. Dysplastic changes can be noted within the juvenile polyps on occasion [9].

JPS is characterized by an autosomal dominant inheritance pattern with variable penetrance; 40–70% of cases are familial, with the remainder occurring sporadically. JPS has been associated with germline mutations in three genes [*SMAD4*, *BMPR1A* (bone morphogenic protein receptor 1A), and *ENG*], all of which are part of the transforming growth factor-β signaling pathway [11]. The role of the *PTEN* gene mutation currently remains controversial, but this mutation can occur in conjunction with mutations of BMPR1A in patients with the rarest form juvenile polyposis of infancy, which is also associated with the highest morbidity and mortality. In the majority of cases, mutation of PTEN likely represents Cowden syndrome or BRRS in patients who have not yet shown the associated extraintestinal clinical features of these conditions rather than JPS.

JPS is associated with an increased lifetime risk of colorectal carcinoma (CRC), thought to arise from adenomatous change within the hamartomas. The incidence of CRC is up to 20% in one study, with a mean age of 34–47 years based on the series and an estimated cumulative risk of 68% by age 60 [9,12▪▪,13]. Gastric cancers may also occur, typically at an older age than CRC. Patients with the SMAD4 mutation are especially likely to have a high gastric polyp burden over time, and in addition to regular endoscopic surveillance, complete or partial gastrectomy may be required in adulthood in some patients [9,12▪▪].

Affected individuals with JPS should undergo surveillance according to current guidelines that include colonoscopy with random biopsies of flat mucosa and endoscopic removal of polyps, if feasible, every 2 years, or earlier based on symptoms. Surveillance esophagogastroduodenoscopy (EGD) should also be started in the early-mid teens; in part, the surveillance interval may be dictated by the genotype if known [9,12▪▪].

In patients with dysplasia, cancer, or a high polyp burden whose symptoms cannot be adequately resolved endoscopically, and in those at high risk for repetitive endoscopic procedures or who may be unreliable for screening, a colectomy with ileorectal anastomosis (IRA) or ileal pouch anal anastomosis is warranted, although infrequently required in childhood. If the rectum is left in place in the case of an ileorectal anastomosis, continued lifelong surveillance of the rectum is required. Asymptomatic first-degree relatives of patients with JPS should be screened for the disease, as they are at risk for juvenile polyposis and colorectal cancer.

JPS–hereditary hemorrhagic telangiectasia overlap syndrome has recently been reported and can occur especially in patients with the SMAD4 mutation [9,11]. In addition to surveillance for JPS, patients with this syndrome should undergo screening/precautions for the vascular malformations associated with hereditary hemorrhagic telangiectasia that include mucocutaneous telangiectasia, epistaxis, and ateriovenous malformations of the lungs, liver, and brain [11].

PTEN hamartoma tumor syndrome includes both Cowden syndrome and BRRS caused by a *PTEN* gene mutation and are inherited in an autosomal dominant pattern or occur as isolated cases [14–16]. Cowden syndrome and BRRS appear to be allelic with the same mutation occurring in patients with both the conditions. Therefore, phenotype may not be predicted by genotype [14]. PTEN is a tumor suppressor gene mapped to chromosome 10q23.31 and functions to cause gastrointestinal cell cycle arrest and/or apoptosis and inhibits cell migration. Decreased expression in cases of mutation is associated with unregulated cell proliferation [14]. In patients with these conditions, polyps resemble juvenile polyps macroscopically and histologically. Patients with

Cowden syndrome and BRRS, however, have extraintestinal manifestations that distinguish them from JPS. Extraintestinal manifestations of Cowden syndrome include mucocutaneous lesions (such as facial trichilemmomas), papillomatous papules, and acral keratoses, as well as other extraintestinal manifestations such as macrocephaly (with normal ventricle size), and thyroid, endometrial, and breast manifestations, which can include cancer [17]. Extraintestinal manifestations of BRRS syndrome, conversely, include developmental delay, macrocephaly, lipomas, and genital pigmentation [18]. Unlike Cowden syndrome, there are no formal diagnostic criteria for BRRS because of the scarcity of this syndrome, but should be considered if there is a family history of BRRS or Cowden syndrome and if a patient has one or more associated extraintestinal manifestations.

Peutz–Jeghers syndrome

PJS is a rare hamartomatous polyposis syndrome (prevalence of one in 50 000–200 000 live births) associated with an increased risk of gastrointestinal and extraintestinal cancers. The lifetime risk of cancer is 37–93%. The most common is CRC, followed by breast, small bowel, gastric, and pancreatic cancers as well as gynecological, lung, and esophageal cancer [19]. PJS is characterized by mucocutaneous pigmentation (small, 1–5 mm pigmented macules, particularly of the lips) and hamartomatous polyps throughout the gastrointestinal tract, most commonly in the small intestine with the highest frequency in the jejunum and less often polyps found in the stomach and colon [20].

The polyps may result in bleeding and anemia, but the highest morbidity is because of the repetitive small bowel intussusception due to large polyps, resulting in intestinal obstruction, and frequently surgery that may result in several bowel resections. The average age of onset of symptoms is in early twenties, but this condition can present in childhood. Diagnosis of PJS can be made when one of the following is present: two or more histologically confirmed PJS polyps, any number of PJS polyps along with characteristic mucocutaneous pigmentation, any number of PJS polyps if there is a family history of PJS in close relative, or characteristic mucocutaneous pigmentation along with a family history of PJS in close relative.

On physical examination, the mucosal pigmentation of the lips is the most characteristic feature, but pigmentation can occur in other areas as well (buccal mucosa, skin around the eyes, nostrils, hands and feet, and perianal region). This pigmentation may start to appear in the first year of life, but may fade during puberty and adulthood. Endoscopically, a PJS polyp is typically a pedunculated polyp with a long stalk. In distinction to juvenile polyps, there is hyperplasia of the smooth muscle layer noted on histology in a characteristic arborizing pattern.

PJS is inherited in an autosomal dominant pattern, with both familial and sporadic transmission with the germline mutation identified as the *STK11/LKB1* gene on chromosome 19p13.3, as the molecular cause in 70% of PJS families and in 50% of sporadic PJS patients. *STK11/LKB1* is a tumor suppressor gene that has an important role in G1 cell cycle arrest, p53-dependent apoptosis, cell polarity, and cellular energy levels [21,22].

Endoscopic surveillance should include an upper endoscopy and colonoscopy starting at 8 years of age, or earlier if symptomatic. If polyps are noted, the examinations should be repeated every 2–3 years. Examination of the small bowel is also needed, by various modalities such as video capsule endoscopy (VCE), magnetic resonance enterography, or small bowel follow through (SBFT) [23]. The location and sizes of the polyps will influence their management. Endoscopic or surgical resection of polyps does not lower the cancer risk in this condition, is not curative or always possible, and it is performed primarily for complications or to avoid complication of PJS polyps.

Owing to the risk of recurrent bowel obstructions necessitating surgery in this condition, strategies have evolved to identify and, subsequently, treat larger small bowel polyps (those $\geq$10–15 mm in diameter) and remove them in a less-invasive manner than surgery in the acute setting. Initially, SBFT was used to try and characterize the polyp burden and determine if large polyps could be removed endoscopically or at the time of intraoperative endoscopy. A recent small pediatric series compared VCE to SBFT examination in terms of detection of larger ($\geq$10 mm) small bowel polyps. In this single-center study from England over a period of 4 years of 11 children with a mean age of 11.2 years, only three patients (27.3%) had large polyps detected and there was poor correlation between SBFT, VCE, and intraoperative findings [24]. However, nonsignificant small bowel polyps were identified in 81.2% of these patients, with VCE having a much higher yield in identifying these polyps than SBFT.

At present, VCE, DBE, or SBE (double or single-balloon enteroscopy), push enteroscopy, computerized tomographic enterography, and magnetic resonance enterography are all utilized to assess the 'polyp burden' and may allow earlier detection of polyps in PJS and, in the case of enteroscopy, allow for nonoperative removal especially of 'at-risk' polyps [12▪▪].

Family adenomatous polyposis

FAP is the most common of the polyposis syndromes in childhood, occurring in one in 10 000 births, and inherited in an autosomal dominant manner, although spontaneous mutations occur in 20–30% of cases. There are several variants of FAP with different genetic mutations associated and varying age of onset as well as age of CRC risk. Classic FAP is diagnosed in patients who ultimately may have more than 100 adenomatous colorectal polyps; patients with attenuated FAP may have fewer polyps and a later age of onset. Patients with severe FAP phenotype may have an earlier age of onset of polyposis and a significantly shorter survival compared with both the classic and attenuated form [25]. There are other FAP variants, including Turcot syndrome (association with brain tumors such as medulloblastoma) and Gardner's syndrome (association with extraintestinal manifestations). Patients with classic FAP may ultimately have hundreds to thousands of adenomatous colonic polyps. These polyps start in childhood or adolescence and increase in quantity with age. Patients with FAP may be surprisingly asymptomatic given the large number of polyps, but symptoms including stools with blood or mucous and diarrhea may occur even in the first decade of life. By the fifth decade of life, progression to neoplasia is almost universal in patients with FAP who have not undergone colectomy. Attenuated FAP has a later presentation as compared with classic FAP but with the same CRC risk [25].

Physical examination is often nondiagnostic in children with FAP unless there is evidence of rectal bleeding or hypoproteinemia. Extraintestinal manifestations including epidermoid cysts, desmoid tumors, fibromas, lipomas, and osteomas (especially mandibular) suggest a variant of FAP formerly known as Gardner's syndrome. At colonoscopy numerous adenomatous polyps can be seen which are small, nodular, and typically sessile, and of variable size in distinction to lymphonodular hyperplasia of the colon (Fig. 2). On histology, the polyps can be tubular, tubulovillous, and villous adenomas with dysplasia ranging from low to high grade. Polyps involving the entire colon and a polyp burden of more than 50 polyps at the time of diagnosis was found in the majority of patients in a recent large pediatric series [26▪▪].

FAP is associated with a germline mutation in the adenomatous polyposis coli (*APC*) gene, located on chromosome 5q21. In 20–30% of cases, however, the mutation is spontaneous with no familial association [20]. The *APC* gene is thought to be a tumor suppressor gene, and also thought to play a role in a variety of cellular functions, including differentiation, apoptosis, and proliferation. There are more than 400 mutations of the *APC* gene described in FAP, and localization of the mutation within the gene correlates with clinical phenotype and severity of clinical manifestations. In a recent pediatric report, the familial pattern of occurrence correlated well with both age of onset and disease manifestations [26▪▪]. Patients identified as a result of a positive family history were diagnosed an average of 2.6 years earlier. Because significant complications including high-grade dysplasia, invasive cancer, and papillary thyroid cancer can occur even in adolescence, early diagnosis and initiation of screening is key, and has been associated with improved survival [25].

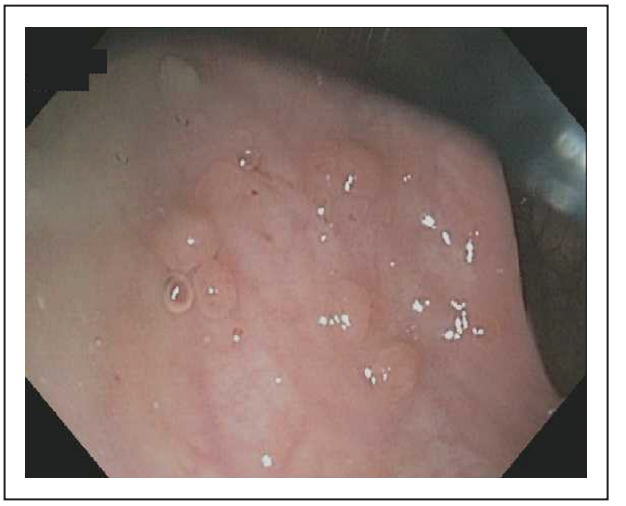

FIGURE 2. Multiple small adenomatous polyps in a teenager with familial adenomatous polyposis. The polyps are irregular in size and distribution in distinction to lymphonodular hyperplasia of the colon that is more uniform in appearance.

Management is based on the patient age and reason for evaluation (presence of clinical symptoms, screening because of an affected family member, etc.). In those families with a known mutation in the index case, genetic screening of primary relatives should start around age 10, younger in families with early-onset phenotype or according to current guidelines [12▪▪,26▪▪]. Those who are gene positive should undergo screening colonoscopy beginning at age of 10–12 years or sooner, with repeat colonoscopies every 1–2 years, in addition to evaluation by a genetic counselor. Those patients who are gene test negative in a family with an identified *APC* gene mutation have the same colorectal cancer risk as the general population, and do not need to undergo special screening colonoscopy but should instead follow standard recommendations for CRC screening. In those families in which there is no known mutation found in the

index case, genetic testing is not informative. These patients will need to undergo regular colonoscopic/endoscopic screening analogous to those individuals that are gene positive.

FAP is confirmed by finding adenomas on colonoscopy with determination of polyp location and density. Prophylactic proctocolectomy is required and is the only therapy that eliminates the inevitable risk of CRC. The timing of colectomy is more challenging in pediatric patients and current guidelines should be followed [12▪▪]. If high-grade dysplasia is absent and patients are asymptomatic, colectomy is usually performed in the mid-late teens or early twenties at the latest. EGD starting in the early twenties and regularly thereafter should be performed, as gastric and duodenal polyps, as well as duodenal ampullary carcinoma, the most second most common cause of death in FAP patients and the most common cause in those who have undergone colectomy, can develop. Some centers start upper tract screening at age 10–12 [12▪▪,26▪▪,27]. Other extracolonic malignancies to screen for include follicular or papillary thyroid cancer starting as a teenager, childhood hepatoblastoma (children up to age 6), and tumors of the central nervous system [12▪▪,26▪▪]. Desmoid tumors, most commonly associated with mutations in the 3′ region of codon 1500 despite being 'benign', can be associated with reduced patient survival because of complications of their growth. The incidence of desmoids is significantly higher in patients who have undergone surgery [25].

Hereditary nonpolyposis colorectal cancer

Individuals later diagnosed with HNPCC can initially present as adolescents with an adenoma found on colonoscopy. Adenomas may be more proximally located or flat in appearance than non-HNPCC adenomas [12▪▪]. HNPCC is inherited in an autosomal dominant manner and is due to a germline mutation in the DNA mismatch repair genes [12▪▪,28]. Patients with this condition have a substantially higher lifetime risk of CRC than the general population as well as an increased risk of synchronous or metachronous CRC and extraintestinal cancers including endometrial and ovarian cancers; current screening guidelines should be followed [12▪▪]. Adenomas in HNPCC have a substantially more rapid adenoma to carcinoma sequence than that which occurs in patients with sporadic cancers; therefore, patients require frequent colonoscopic screening and may ultimately require colectomy with ileal pouch anal anastomosis [12▪▪]. The colonic tumors in patients with HNPCC have associated changes in the length of nucleotide sequences of tumor DNA termed microsatellite instability that can be detected in laboratory testing of the tumor specimens, and may be the initial clue to identification of HNPCC in a kindred.

POLYPOID NEOPLASMS OF THE COLON

Gastrointestinal neoplasms, both primary and metastatic, are uncommon and may not be recognized in children, having the appearance of an irregular polyp or mass on colonoscopy. Presenting symptoms can be variable and include abdominal pain, distension, weight loss, vomiting, a palpable mass, and gastrointestinal bleeding. Endoscopic polypectomy of colonic tumor may be extremely hazardous and associated with an increased risk of significant complications including bleeding and perforation, as tumors erode through the bowel wall. Therefore, endoscopic biopsy is a safer initial step if a polypoid lesion suspicious for cancer is encountered at colonoscopy in a child.

CONCLUSION

Pediatric colonic polyps are common in young pediatric patients, usually are solitary, and the vast majority is benign with no premalignant potential. Young children with multiple polyps (synchronous or metachronous), adolescents with juvenile polyps, and any child with an adenomatous polyp (single or multiple) or polyps in the stomach or small bowel should be evaluated for a polyposis syndrome that includes a careful family history for gastrointestinal and extraintestinal malignancies. Depending on the specific polyposis syndrome, there are significant implications for endoscopic surveillance, colectomy in some cases, as well as additional cancer screening based on current guidelines.

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Conflicts of interest

There are no conflicts of interest.

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- ▪ of special interest
- ▪▪ of outstanding interest

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