APC-Associated Polyposis Conditions

Record Status
This is a bibliographic record of a published health technology assessment. No evaluation of the quality of this assessment has been made for the HTA database.

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Authors' objectives
Colorectal cancer (CRC) is the third most common cancer in both men and women in the United States, affecting nearly 150,000 Americans each year and causing more than 50,000 deaths annually. The majority of CRC is sporadic in nature, but approximately 15% of patients have a family history of cancer. APC-associated polyposis syndromes are a collection of genetic CRC syndromes that are caused by sequence variants in the adenomatous polyposis coli (APC) gene, a tumor suppressor gene located on chromosome 5 at bands q21 to q22. These conditions include familial adenomatous polyposis (FAP) and its phenotypic variants Gardner's syndrome and Turcot's syndrome, as well as the milder attenuated familial adenomatous polyposis (AFAP). APC-associated polyposis syndromes account for less than 1% of CRC. FAP is characterized by the presence of hundreds to thousands of adenomas in the colon and rectum. Approximately half of all FAP patients develop adenomas by 15 years of age, with nearly 95% of patients developing adenomas by their mid 30s, leading to an almost 100% chance of malignant transformation in at least one of these polyps by the fifth decade of life. In addition to the characteristic polyp formation in the colon, individuals with FAP also have a significant risk of both extracolonic gastrointestinal and extraintestinal manifestations. Polyps may develop in the upper gastrointestinal tract, especially the duodenum. Other manifestations of FAP that are variable between individuals are gastric fundic polyps, small bowel (especially duodenal) adenomatous polyps, congenital hypertrophy of the retinal pigment epithelium (CHRPE; patches of discoloration in the ocular fundus), supernumerary teeth, osteomas and cutaneous lipomas, desmoids, and cysts. Gardner syndrome is characterized by the features of classic FAP with the occurrence of soft-tissue tumors (sebaceous cysts, lipomas, desmoid tumors, and fibromas). Turcot's syndrome is a rare disorder characterized by central nervous system (CNS) tumors occurring together with colonic polyposis. AFAP is a milder phenotypic variant of FAP that is characterized by fewer polyps that appear approximately 10 years later than with FAP; typically, < 100 colonic polyps are diagnosed at a mean age of 41 years. The risk of CRC appears to be lower in AFAP, but when it does occur, the median age at diagnosis is 55 years. The most common treatment approach to prevent FAP is a total proctocolectomy with ileostomy, although subtotal colectomy with ileorectal anastomosis (IRA) and proctocolectomy with ileal pouch-anal anastomosis (IPAA) may also be considered.

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