

Familial Adenomatous Polyposis within the Pediatric Hereditary Polyposis Registry

Brendan Anderson¹, Tina Zhang²

¹Indiana University School of Medicine; ²Indiana University School of Medicine, Riley Children's Hospital Department of Pediatric Gastroenterology

Background/Objective:

Familial Adenomatous Polyposis (FAP) is a condition characterized by values exceeding hundreds of colorectal adenomatous polyps. FAP has a prevalence of 1 in 8,300 people, demonstrating an autosomal dominant inheritance pattern. This condition follows a pathogenic variation of the Adenomatous Polyposis Coli (APC) gene, located on chromosome 5q21-22. These mutations are heavily correlated with the incidence of colorectal carcinoma, requiring prophylactic colectomy. The management and treatment of FAP requires routine surveillance to reduce gastrointestinal polyp burden. The objective for this study is to characterize the current patient population at Riley with polyposis syndromes and assess clinical surveillance in patients diagnosed with FAP.

Methods:

Historical data from 2020-2022 GI clinical visits were reviewed using the diagnostic codes *Family history of colonic polyps*, *Benign neoplasm of colon unspecified*, and *Other phakomatoses not elsewhere classified*. These criteria identified 93 patients of which 64 patients were diagnosed with a hereditary polyposis syndrome for inclusion within the registry. Of the 64 patients, 42 had a diagnosis of FAP. Clinical data reviewed included the patient's age of diagnosis, completion of genetic testing and follow-up, surgical treatment, routine endoscopic surveillance, and modes of imaging.

Results:

Results indicated a median age of diagnosis of 10 years, lower than previously reported national averages of 13.5-17 years. Evaluation of routine follow-up indicated a rate of endoscopy at 0.822 scopes per year and demonstrated that abdominal CT scans as well as abdominal X-rays were the most common supplemental modes of imaging.

Conclusion/ Future Directions:

The data from this registry will help direct care of patients with FAP, ensuring they receive treatment in accordance with national guidelines for routine surveillance and prophylactic colectomy. Moving forward, the intention is to expand the registry's date range to incorporate more patients and elaborate further on endoscopic findings for the specific subtypes of FAP.