Chapter 10

GASTROINTESTINAL POLYPOSIS SYNDROMES

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INTRODUCTION

Gastrointestinal (GI) polyposis syndromes are very common lesions within GI system with multiple phenotypes (Table 1). GI polyp subtypes can be associated with hereditary polyposis syndromes and link to colon cancer. Hence, polyps of the colon are distinctly different than GI polyposis syndromes both pathologically and clinically.

INHERITED POLYPOSIS SYNDROMES

Inherited syndromes represent phenotypes of disorders with distinct inheritance patterns and multiple polyps in the colon.

Familial Adenomatous Polyposis (FAP)

Adenoma is the most common form of polyp seen in individuals with FAP and is considered a pre-cancerous polyp and if not treated an increased risk for hereditary colorectal cancer (CRC) syndrome occurs. FAP is the second common hereditary sydrome after hereditary non-polyposis colorectal cancer syndrome (HNPCC; Lynch syndrome) that causes colon cancer if not treated. FAP is an autosomal dominant disorder (80-100% penetrance) (Feldman,M. etal. (2015). and has an estimated prevalence of 1 in 10000 live births (Jarvinen HJ et al (1992), Bisgaard ML et al (1994), Bulow S et al (1996)). People with the classic type of FAP may begin develop polyps as early their teenage years and could develop 100 to 1,000 colon polyps. Individuals with FAP are also at risk for developing cancer in other organs such as duodenum, stomach, pancreas, hepatoblastoma, thyroid, osteomas and desmoid tumors.

There are different types of adenomatous polyps based on the growth patterns i.e. tubular, villous or a mixer of the two called tubulovillous adenomas. Most polyps are small (less than 1 cm). If FAP is not treated, the likelihood that CRC develop is very high. Colonoscopy is recommended every year beginning at the age 10-12, to minimize the risk of CRC development.

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