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Cancer within the family tree: risks, diagnosis and treatment of juvenile polyposis syndrome

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DESCRIPTION

A 41-year-old woman with irritable bowel syndrome symptoms presented for colonoscopy after her mother was diagnosed with multiple juvenile colon polyps positive for *Bone morphogenic protein receptor type-1A* (*BMPR1A*) mutation and subsequently colon cancer at age 64. Colonoscopy revealed multiple irregular pedunculated polyps (1–4 cm), including the tubular adenoma in figure 1, and a circumferential, broad-based 4–5 cm tubulovillous adenoma seen in figure 2 that nearly completely obstructed the mid-ascending colon. Biopsies revealed tubular adenomas and tubular villous adenomas with positive *BMPR1A* mutation suggesting juvenile polyposis syndrome (JPS).

JPS is clinically diagnosed in the absence of other hamartomatous polyposis syndromes, including Peutz-Jeghers and Cowden with the presence of either greater than five juvenile polyps in the colorectum, multiple juvenile polyps elsewhere in GI tract or any polyps with family history of juvenile polyps.[1 2] Presence of JPS confers a cumulative colorectal cancer risk of 68% by age 60 years.[2]

BMPR1A is an autosomal dominant gene mutation that affects the transforming growth factor-beta pathway leading to hundreds of polyps often within the first decade of life. Mutations are found in nearly 60% of JPS cases.[2]

At-risk JPS patients imperatively need genetic testing for both *SMAD4* and *BMPR1A* gene mutations to determine appropriate cancer screening. Current European Society of Gastrointestinal Endoscopy guidelines suggest early screening of the stomach at age 18 years and colon as early as age 12 years, as the single most impacting factor in outcomes of patients and their families.[3] Our patient was referred to general surgery for resection of mass.

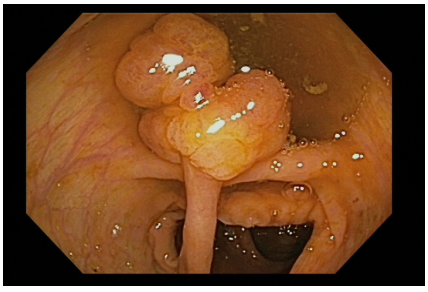

Figure 1 Tubular adenoma found in the mid ascending colon.

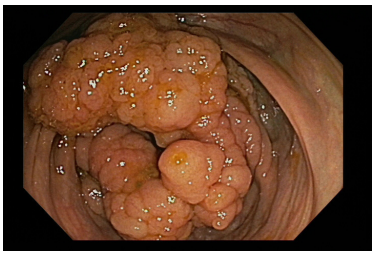

Figure 2 Nearly obstructing 4–5 cm tubulovillous adenoma in mid-ascending colon.

Contributors All persons who meet authorship criteria are listed as authors, and all authors certify that they have participated sufficiently in the work to take public responsibility for the content, including participation in patient care, analysis, writing, and revision of the manuscript. PM was main physician responsible for direct patient care while KK and RLS were main writers of the manuscript. MA was involved with significant research and editorial assistance. Furthermore, each author certifies that this material or similar material has not been and will not be submitted to or published in any other publication before its appearance in the Postgraduate Medical Journal BMJ.

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Learning points

- Juvenile polyposis syndrome (JPS) is clinically diagnosed in the absence of other hamartomatous polyposis syndromes with the presence of either greater than five juvenile polyps in the colorectum, multiple juvenile polyps elsewhere in the gastrointestinal tract or any polyps with family history of juvenile polyps.
- JPS confers a cumulative colorectal cancer risk of 68% by age 60 years.
- European Society of Gastrointestinal Endoscopy guidelines recommend patients with JPS to undergo early screening of colon as early as age 12 years and stomach at age 18 years via Esophagogastroduodenoscopy.

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