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Hyperplastic polyposis syndrome: a call for broader diagnostic criteria

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Background

Hyperplastic Polyposis Syndrome (HPS) is a rare disease characterized by multiple or large hyperplastic polyps and carries an approximately 40% lifetime colorectal cancer risk. Although a genetic basis has not been established, HPS is believed to be a heritable syndrome and is diagnosed by clinical criteria as set forth by the World Health Organization (WHO). Based on clinical experience, we hypothesized that WHO criteria may be narrowly restrictive and misses some patients with an increased malignancy risk.

Methods

For this study, HPS was defined by meeting at least one of the following criteria: 1) ≥20 HPs anywhere in the colon, 2) ≥5 HPs proximal to the sigmoid colon, 3) ≥2 HPs at least 10mm in size, 4) any HPs and a 1st degree relative with HPS. Colonoscopy and pathology databases were retrospectively reviewed for patients meeting criteria. Patient demographics, colonoscopic findings, and personal and family history of cancer were recorded.

Results

Sixty patients (38 males, 22 females) meeting at least one of the above criteria were included. Only 19 of these 60 patients (32%) also satisfied WHO criteria for HPS. Results are summarized in Table 1. Importantly, of the additional 41 patients only meeting the broader criteria, 27% had a personal history and 44% had family history of colorectal cancer. This group also had extracolonic malignancies including breast, lung, prostate, and testicular cancer.

Table 1

HPS Criteria	N	Colorectal Cancer History		Other Cancer History	
		Personal	Family	Personal	Family
WHO	19	5 (26%)	4 (21%)	8 (42%)	7 (37%)
Only New Criteria	41	11 (27%)	18 (44%)	9 (22%)	11 (29%)
All Patients	60	16 (27%)	18 (37%)	12 (23%)	18 (30%)

Conclusion

HPS is associated with a personal and familial risk of colorectal cancer and other malignancies. Applying broader definitions identifies an additional population of patients with increased personal and familial cancer risk. More inclusive criteria should be used until a genetic basis of disease better defines cancer risk.

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