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Clinicopathologic and genetic features of young patients with colorectal cancer

Melyssa Aronson*, Steve Gallinger, Heidi Rothenmund, Kara Semotiuk, Spring Holter, Terri Berk, Aaron Pollett, Zane Cohen, Bharati Bapat, Hyeja Kim, Robert Gryfe

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Background

Colorectal cancer (CRC) is the 3rd most common cancer in Canada, often occurring at older ages. While early-onset CRC can be suggestive of an inherited syndrome, the underlying genetic cause remains unexplained in many of these young individuals.

Method

Clinicopathologic features along with genetic and family information were collected on individuals diagnosed with CRC ≤35 years old identified through the Familial Gastrointestinal Cancer Registry in Toronto, Canada.

Results

441 individuals from 353 families were identified, and to-date, medical records confirmed 254 diagnoses, which were included for analysis. Ninety patients (35.4%) had germline mutations in self or kin (31 *MSH2,* 36 *MLHI,* 1 *MSH6,* 3 *PMS2,* 24 *APC,* 2 *MYH biallelic* and 1 *BRCA2).* Individuals were classified into six categories; (a) 74 had Lynch syndrome (LS) confirmed by germline mutation or tumour deficiency (b) 4 had constitutional mismatch repair-deficiency (CMMR-D) (c) 61 had polyposis (mutation positive, >25 adenomas or hamartomatous polyps), (d) 6 met Family X criteria, (e) 7 had inflammatory bowel disease (IBD), and (f) 102 were unclassified (NOS), with 69 of these individuals having MSS and/or IHC intact tumours.

On average, patients with CMMR-D presented younger, with a mean age of 14 years old at diagnosis. 65.2% of patients with LS presented with a proximal tumour (65.2%) compared with the polyposis (10.2%) and NOS (34.4%) groups. In contrast, 83% of polyposis patients and 65.5% of NOS patients presented with distal colon or rectal cancers.

Family history was significant for the majority of LS patients with 54 of 73 (74%) meeting Amsterdam I or II criteria. Two of the 7 patients with IBD, and 15 of 98 NOS patients also met Amsterdam *I/II* criteria, as opposed to the CMMR-D families where none met these criteria. Six LS patients presented with sporadic CRC, as did approximately 25% of polyposis patients and 59.2% of the NOS patients.

Conclusions

Cancer site differs between individuals ≤35 years with LS, polyposis and NOS CRC. Greater than 1/3 of patients presented with no significant family history of CRC. While the majority of patients diagnosed with CRC ≤35 have a known hereditary CRC syndrome or risk predisposition, 69 of 221 (31.2%) of individuals appear to have no recognizable syndrome.

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* Correspondence: maronson@mtsinai.on.ca
Dr. Zane Cohen Digestive Disease Clinical Research Centre, Mt. Sinai Hospital, Toronto, Ontario, Canada