

POSTER PRESENTATION

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# Clinicopathologic and genetic features of young patients with colorectal cancer

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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  $\leq 35$  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 *MLH1*, 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  $\leq 35$  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  $\leq 35$  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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