Descriptive Analyses of Patients with Familial Adenomatous Polyposis at Hospital de Câncer de Barretos

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Introduction: Familial adenomatous polyposis (FAP) is generally caused by germline mutation in *APC* gene, and is characterized by adenomatous polip growth in the gastrintestinal tract. FAP is responsible for around 1% of colorectal cancer (CRC) incidence and also is related to duodenal adenocarcinoma and desmoids tumors. At Barretos Cancer Hospital (BCH), patients with adenomatous poliposis (≥ 10 adenomas) are referred to Oncogenetics Department for genetic counseling. All services (including genetic testing) are free of charge for patients and their family. Objectives: To describe clinical presentation and management of FAP patients from BCH. Methodology: Retrospective case series report reviewed from BCH family records, between September/2009 and December/2015. Surveillance was performed according to NCCN guidelines. Results: We have evaluated 152 individuals who tested positive, from 48 families. Probands median age at the diagnosis were 31 years old (range 13y-49y). The relatives median age was 27 years old (range 12y-66y). Between the probands, 22 (45.8%) already had a previous diagnosis of CRC, one (2%) had ileal adenocarcinoma and one had gastric cancer. Between the relatives who underwent genetic test, 15 (14.4%) had CRC previously, one had (0.9%) gastric cancer and one had pancreatic adenocarcinoma. Semestral colonoscopic screening is performed in those who has mild colorectal polip burden (18 individuals between ages 13 and 39. The most common mutations detected in *APC* gene were: c.874C>T (8.3% of the families), c.904C>T (8.3% of the families), c.3183_3187delACAAA (8.3% of the families), c.3927_3931delAAAGA (8.3% of the families). Conclusion: Considering the short period assessed, the large coverage area BCH supports and the perspective of expanding the Service, these preliminary results could be a source for better characterization for LS in Brazilian families.