Descriptive Analyses of Lynch Syndrome Carriers at Barretos Cancer Hospital

Henrique C. R. Galvão (Barretos' Cancer Hospital, Brazil), Cristina Sabato (Barretos' Cancer Hospital, Brazil), André E. de Paula (Barretos' Cancer Hospital, Brazil), Maximiliano C. Neto (Barretos' Cancer Hospital, Brazil), Carlos E. Paiva (Barretos' Cancer Hospital, Brazil), Luis G. C. Romagnolo (Barretos' Cancer Hospital, Brazil), Edenir I. Palmero (Barretos' Cancer Hospital, Brazil), Rui M. Reis (Barretos' Cancer Hospital, Brazil), Edmundo Mauad (Barretos' Cancer Hospital, Brazil)

Introduction: Up to 5% of colorectal cancer (CRC) cases are due by Lynch syndrome (LS), caused mainly by mutations in the mismatch repair (MMR) genes *MLH1, MSH2, MSH6* and *PMS2.* At Barretos Cancer Hospital (BCH), patients who meet criteria are referred to Oncogenetics Department for genetic counseling. All services (including genetic testing) are free of charge for patients and their family. Objective: To describe clinical presentation and management of LS carriers from BCH. Methodology: Retrospective case series report reviewed from BCH family records, between April/2010 and December/2015. Surveillance was performed according to NCCN guidelines. Results: We evaluated 144 individuals who tested positive, from 51 families (39,2% *MLH1*, 35,3% *MSH2*, 15,7% *MSH6* and 9,8% *PMS2*). Probands median age at the diagnosis were 51 years old (range 11y-76y). The relatives median age was 39 years old (range 18y-74y). All 51 probands had at least one previous diagnosis of cancer: 48 CRC cases, 17 endometrial cancer (EC) cases, four gastric cancer cases and two ovarian cancer cases. The median age at CRC diagnosis was 42 years old and 47 years old for EC. Between the 93 relatives that were tested positive, 20 already had a previous diagnosis of cancer: 10 CRC, five EC, two urothelial and one prostate cancer. In 73 asymptomatic carriers (individuals with a positive predictive test), we had detected six cases of malignant neoplasia during a median surveillance period of 16 months. The most common mutations detected were: c.1276C>T (p.Gln426Ter) in *MLH1* (11,8% of the families) and three large deletion between exons 17 and 19 in *MLH1* (5,9% 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.