Descriptive Analyses of *BRCA1/2* Mutation Carriers at Hospital de Câncer de Barretos

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Background: Germline mutations in *BRCA1* or *BRCA2* genes are related to 2%-3% of breast cancer (BC) cases and up to 15% of ovarian carcinoma (OC). Genetic counseling is required for individuals facing this condition. Barretos Cancer Hospital (BCH) offer genetic counseling and genetic testing free of charge for patients and their families. Objective: To describe clinical presentation and management of *BRCA1/2* mutation carriers from BCH. Methodology: Retrospective case series report reviewed from BCH family records, between May/2010 and December/2015. Surveillance was performed according to NCCN guidelines. Results: We have assessed 158 individuals carrying *BRCA1* and 106 carrying *BRCA2* mutation from 109 families (66,9% *BRCA1* mutated). Probands median age (all female) at the time syndromic diagnosis was confirmed was 48 years (range 24y-77y). The relatives median age was 42 years (range 18y-82y). All, except one, of the 109 probands had at least one previous cancer already diagnosed. There were 23 cases of primary OC and 110 cases of BC among those patients (41,8% triple negative BC, all linked to *BRCA1* mutations, except for three of them). The median age at BC diagnosis was 40 years and it was 53 years for OC diagnosis. Of those 154 relatives who tested positive (70,1% female), 24 already had cancer previously (19 BC, one OC and two had other types). Between 130 asymptomatic carriers (individuals with a positive predictive test), we had detected three case of BC and one case of OC during an average surveillance period of 16 months. The most common mutations detected were: c.5266dupC in *BRCA1* (24,5% of the families), p.Gln1111Asnfs (c.3331_3334delCAAG) in *BRCA1* (10,9% of the families) and c.2808_2811delACAA in *BRCA2* (3,6%). Conclusion: Considering the period assessed, the large coverage area BCH supports and the perspective of expanding the Service, these results could be a source for better characterization for hereditary breast/ovarian cancer predisposition syndrome in Brazilian families.