PROFILE OF PATIENTS WITH BREAST AND / OR OVARIAN CANCER ATTENDED AT THE INCA ONCOGENETICS AMBULATORY AND EVALUATION OF THE PENN II PREDICTION MODEL

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INTRODUCTION

Breast cancer is among the major malignancies that affect women in Brazil and in the world, being considered a global health issue. The estimate of new cases in 2018 is 59.700, with an estimated risk of 56.33 cases per 100.000 women.

Regardless non-melanoma skin tumors, breast cancer is the most frequent in women from South (73.07/100.000), Southeast (69.50/100.000), Midwest (51.96/100.000) and Northeast (40.36/100.000) regions from Brazil. In the North region, it's the second most incident tumor (19.21/100.000).

Despite the decrease in mortality rates, there is an increase in its incidence as well as the mutations related to *BRCA1* and *BRCA2* genes. It's important to identify individuals with pathogenic mutations in these genes in order to follow the case, aiming to reach the best outcome to the patient and their relatives. Taking into account that molecular analysis of all suspected mutations is laborious and not cost-effective, the use of prediction models has become a valuable tool in identifying patients at higher risk of carrying pathogenic mutations in genes related to breast and ovarian cancer.

METHODS

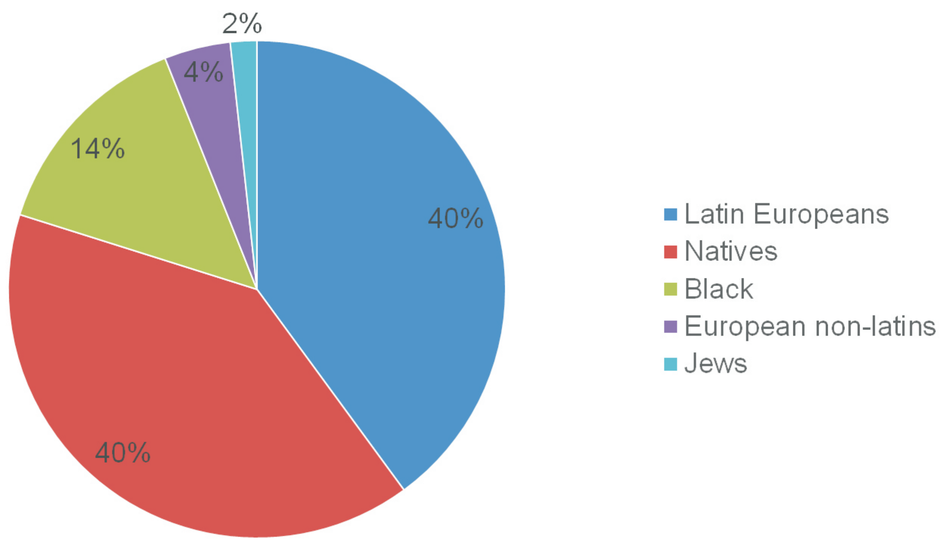
The information of the patients assisted at the Genetic Counseling department between 1997 and 2017 were obtained during the consultations, through a formulary and also from the medical records. Complete sequencing of the *BRCA1/2* genes was performed by the Sanger method and/or by new generation sequencing (NGS). Evaluation of the Penn II prediction model has been performed for all patients in order to access the risk of presenting mutations in *BRCA1/2* genes.

RESULTS

To date, 248 women and 10 men have been analyzed. Graph 1 shows de ancestry of the research subjects according to the self-declaration. Only the most frequent ancestry records were included.

The mean age of the menarche was 12.86 years old (8-20, S.D.: 1.92) and that of the first pregnancy was 22,24 years old (14-39, S.D.: 4,75). Ninety patients (36.3%) took contraceptives between 14 and 40 years old between 1 to 360 months (mean: 72.5 months).

Graph 1: Ancestry of the research subjects



The most frequent histological type of breast cancer was invasive ductal carcinoma present in 158 patients (61.24%). Forty-six had bilateral breast tumors, being 12/46 synchronous and 34/46 metachronous. The age at diagnosis ranged from 16 to 79 years (mean: 44.42, SD: 11.26, median: 44) and in those who presented a second tumor, ranged from 25 to 65 years (mean: 47.21; 10.49, median: 45). Of the patients with breast cancer, four also had ovarian tumor.

Penn II prediction model was performed in order to classify the patients at high and low risk of presenting a pathogenic mutation in *BRCA1/2.* The value adopted to define the two groups was 10%, and patients at high risk were those with Penn II higher than the cutoff value (10%).

Sequencing of *BRCA1/2* in 144 patients (144/258) identified pathogenic mutation in twenty (23.2%) of the high risk group (Penn II > 10%) and in 5 (17.9%) of the low risk group (Penn II < 10%). The difference between these groups was not statiscally significant (Chi-square = 0.35, p = 0,55).

CONCLUSION

In the study group, Penn II did not infer the presence of pathogenic mutation among patients with high or low risk. In order to improve the inference capability of the method, the number of patients included in the analysis will be increased.

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