The impact of family history on breast cancer stage: data from a Brazilian breast cancer cohort without inherited predispositions

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Introduction

Family history (FH) of breast cancer (BC) is an independent risk factor for BC, regardless the presence of a germline mutation in a BC predisposition gene. Women with a FH of BC could be diagnosed at earlier stages due to increased awareness and screening. Nevertheless, other factors could be associated with diagnosis at later stages such as early onset, fear, and misinformation.

Objective

To determine if women with a family history of breast cancer present earlier stages of disease.

Methods and Materials

This single institution study was conducted at the Oncogenetics Department of Hospital Sirio-Libanês (Brasília DF, Brazil). Eligibility included patients with a personal history of BC who received genetic counseling between 2017 and 2025, and a genetic testing result without a pathogenic/probably pathogenic germline variant in a BC gene. Lack of FH data was an exclusion criteria. Data were collected retrospectively from medical charts. FH of BC was defined as a BC cancer diagnosis in 1st, 2nd, and/or 3rd degree relatives (defined as close relatives). For patients who received neoadjuvant therapy, BC stage was based on clinical staging and pathological or clinical staging were used for the remaining cases. Chi-square test was used for comparison of patients with and without a FH of BC.

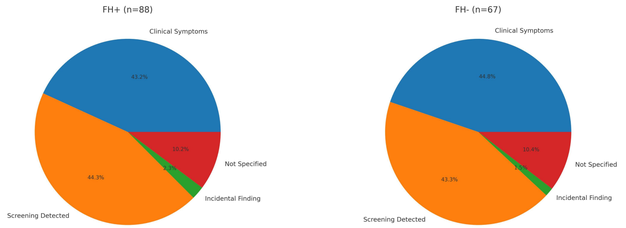
Results

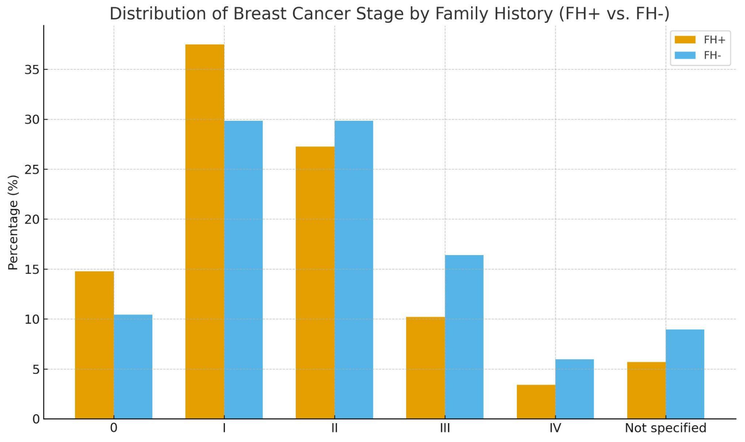
Among 161 patients, 155 met study criteria. Median age of BC diagnosis was 49 years (23.0-87.0), 6 patients were diagnosed during pregnancy or breastfeeding, 6 had bilateral synchronous BC and 7 metastatic de novo disease. Out of 155 patients, 88 (56.7%) had at least one close relative with BC (FH+), including 37 (23.9%) with a family member affected before 50 years and 7 (4.5%) with bilateral BC. In total, 68 (43.8%) patients were diagnosed due to symptoms, 68 (43.8%) through screening, 3 incidentally found during mammoplasty and in 16 cases presentation was not specified. Among those screen-detected cancers (n=68), 22 (32.3%) were detected by ultrasound, 35 (51.5%) by mammogram, 7 (10.3%) by MRI, and in 4 cases the diagnostic modality was unavailable. There were no differences between groups (FH+ vs FH-) regarding age of BC diagnosis, menopausal status, BC presentation, and BC stage.

Table 1. Variables evaluated.

Variable	Category	Overall cohort N=155 (%)	FH+ N=88 (%)	FH- N=67 (%)	P value
Age of BC diagnosis	<50 >/=50	83 (53.54) 72 (46.45)	45 (51.13) 43 (48.86)	38 (56.71) 29 (43.28)	0.598
Menopausal status at BC diagnosis	Premenopausal Peri/Post menopausal Not specified	76 (49.03) 73 (47.09) 6 (3.87)	42 (47.72) 43 (48.86) 3 (3.40)	34 (50.74) 30 (44.77) 3 (4.47)	0.777
BC presentation	Clinical symptoms Screen-detected Incidentally found Not specified	68 (43.87) 68 (43.87) 3 (1.93) 16 (10.32)	38 (43.18) 39 (44.31) 2 (2.27) 9 (10.22)	30 (44.77) 29 (43.3) 1 (1.49) 7 (10.44)	0.927
BC stage	0 I II III IV Not specified	20 (12.90) 53 (34.19) 44 (28.38) 20 (12.90) 7 (4.51) 11 (7.09)	13 (14.77) 33 (37.50) 24 (27.27) 9 (10.20) 3 (3.40) 5 (5.68)	7 (10.44) 20 (29.85) 20 (29.85) 11 (16.41) 4 (5.97) 6 (8.95)	0.562

Across all variables tested, **no statistically significant association** was found with **family history status (FH+ vs FH–)** at the conventional p < 0.05 threshold.





Discussion

Although FH is an independent risk factor for BC, our data, consistent with the non-significant p-value (P=0.562) for the BC stage, suggest that FH alone may not be sufficient to significantly influence the timing of diagnosis in this specific scenario. This can be explained by other factors, such as fear or misinformation, which can lead to diagnosis at later stages, even in the presence of FH.

The lack of difference in the stages diagnosed between the FH+ and FH- groups implies that the isolated use of FH to guide screening in patients without known mutations may be insufficient to optimize early detection.

Conclusions

In this BC Brazilian cohort from a private health setting without a cancer predisposition syndrome, a family history of BC did not impact BC stage at diagnosis. Cancer risk assessment based on risk models such as Tyrer Cuzick could inform the benefit of BC screening with MRI in this scenario.

Contact

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