MALE BREAST CANCER: GENOMIC LANDSCAPE, EPIDEMIOLOGICAL PATTERNS, AND CLINICAL OUTCOMES IN THE UNITED STATES

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INTRODUCTION

- Male breast cancer is a rare form of breast cancer that has its distinct biological characteristics.
- Because of the disease rarity, most data on genomic landscape, epidemiological patterns, and clinical outcomes originate from small studies with limited number of patients.
- In this study, we aimed to leverage the publicly available genomic and epidemiological datasets to provide the largest to date analysis of clinicogenomic characteristics of male breast cancer in the United States.

METHODS

- This is a retrospective study of patients with male breast cancer.
- We utilized the Surveillance, Epidemiology, and End Results (SEER) database to extract data on patients diagnosed with male breast cancer from the SEER 17 Reg. Nov 2024 submission dataset (including patients diagnosed 2000-2022) .
- We used the AACR project GENIE v17.0 database to extract data on genomics of patients with male breast cancer who had samples sequenced and results accessible as part of AACR project GENIE.
- Only patients with one profiled sample were included to avoid duplication.
- Only genes which were sequenced in at least 50 samples were included in frequency calculations.

RESULTS

Epidemiology

- We included a total of 10,284 patients.
- **Race:** Most patients were white (80.3%) and non-Hispanic non-Latino (92.7%).
- **Side:** 51.7% left-sided.
- **Incidence:** The incidence rate was 1.2 per 100,000.
- **Overall survival:** The median overall survival was 109 months (95% CI: 105.1–112.9) (**Figure 1**).

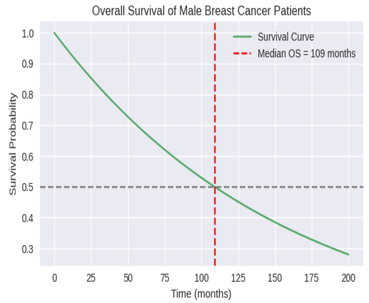


Figure 1. Kaplan–Meier Survival Curve.

Hormonal Status:

- The most common molecular subtype was HR+/HER2– which was identified in 85.9% of patients (**Figure 2**)

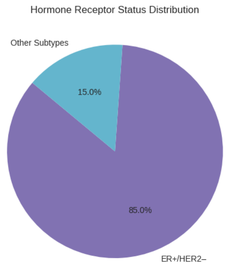


Figure 2. Hormone Receptor Status.

Genomics:

The most frequently mutated genes were:

- *PIK3CA* – 38.2%
- *GATA3* – 21.6%
- *KMT2C* – 14.1%
- *TP53* – 13.9%
- *BRCA2* – 10.4%

The most frequent genes with structural variants included:

- *INPPL1* – 3.4%
- *CDK12* – 1.7%
- *FGFR2* – 1.6%
- *FGFR1* – 1.6%
- *NSD3* – 1.2%

The most common genes with copy number alteration included:

- FGF19 / FGF4 / FGF3 – 19.4%
- CCND1 – 17.8%
- FGFR1 – 14.9%

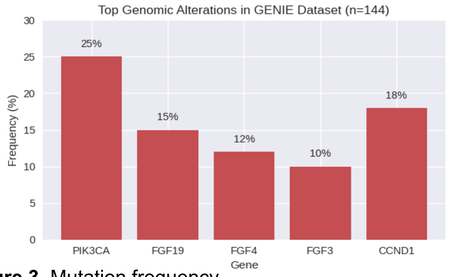


Figure 3. Mutation frequency

CONCLUSION

Male breast cancer occurs at a rate of 1.2 per 100,000 and has a median OS of 9 years. Patients mostly present with ER+ HER- disease. Molecular profile has been revealed.

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