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Knowledge of Tumor/Blood Genomic Testing (NGS) and *ESR1* Mutations in a Survey of Patients with ER+/HER2- Metastatic Breast Cancer (mBC)

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Introduction

- In patients with ER+/HER2- metastatic breast cancer (mBC), mutations in the estrogen receptor-α gene (*ESR1*) acquired from endocrine therapy can lead to treatment resistance, metastasis, and poor prognosis
- Patient awareness of genomic testing and *ESR1* mutations and their potential influence on breast cancer therapy is unknown, as is the impact of patients' demographic characteristics on this knowledge
- A better understanding of this awareness will help oncologists optimize patient education around these increasingly important topics

Objective

To understand awareness of genomic testing and *ESR1* mutations in patients with ER+/HER2- mBC

Methods

- The 42-question, online EQUALS (*ESR1* QUAlity of Life Survey) was sent (June-September 2022) to US patients with mBC from
  - The Cure Media Group (by email)
  - Private Facebook groups of patients with mBC
  - The advanced breast cancer clinic at St. Luke's Cancer Institute
  - FORCE (Facing Hereditary Cancer EMPOWERED)
  - The Chrysalis Initiative
  - METAvivor: mBC research, support and awareness
- Participants were eligible if they had ER+/HER2- mBC
- A $10 gift card was awarded to participants at survey completion
- Survey answers were summarized descriptively

Results

Patient and oncologist characteristics

- 474 patients completed the survey over 4 months
- Participants had a mean age of 45 years (range, 19–83 years), and a mean mBC diagnosis year of 2018; one-third were non-White; 50% had a mean household income <$75K, and 72% had a higher education degree (Table 1)
- Patients' oncologist's gender (female 65%) and practice type (general [56%], breast cancer only [44%]) were well balanced

Prior metastatic breast cancer treatments

- Most common first-line mBC treatments were aromatase inhibitor (AI) alone, AI + CDK4/6 inhibitor (CDK4/6i), fulvestrant + CDK4/6i, and selective estrogen receptor modulator (SERM) (Figure 1)
  - Second-line therapies were fulvestrant + CDK4/6i (28%), AI + CDK4/6i (27%), or AI alone (16%)
- Of the 77% (366/474) who had received chemotherapy in the metastatic setting, 45% (165/366) had received ≥3 lines of chemotherapy

Table 1. Baseline patient characteristics

Characteristics		ER+/HER2- mBC (n=474)
Age, y	Mean ± SD (range)	45 ± 14 (19–83)
Race/ethnicity, n (%)*	White	319 (67)
	Hispanic/Latino	112 (24)
	Black/African American	32 (7)
	American Indian/Alaskan Native	12 (3)
	Asian	7 (1)
	Declined to answer	1 (0)
Living setting, n (%)	Rural	144 (30)
	Suburban	162 (34)
	Urban	168 (35)
Average household income $, n (%)	<25,000	14 (3)
	25,000 to <50,000	116 (25)
	50,000 to <75,000	104 (22)
	75,000 to <100,000	83 (18)
	100,000 to <150,000	87 (18)
	≥150,000	46 (10)
	Decline to answer	24 (5)
Highest level of education, n (%)	Some high school	7 (1)
	High school	125 (28)
	Bachelor's degree	244 (51)
	Master's degree	79 (17)
	Doctoral degree (law, medical, PhD)	19 (4)
mBC diagnosis, y	Mean (range)	2018 (1989–2022)

*Could select more than one option.

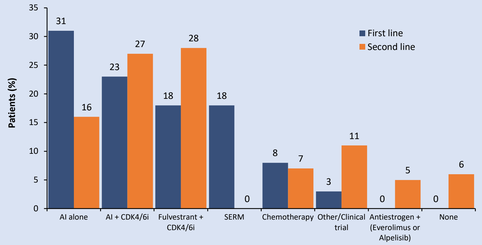
Awareness of liquid biopsies and genomic testing

- When asked about their familiarity with liquid biopsies (circulating tumor DNA assessment from blood), 57% knew a lot/moderate amount (Figure 2A)
  - 50% of patients' oncologists had explained liquid biopsies
- About two-thirds knew a lot/moderate amount about genomic testing (looking at gene expression changes in tumors to help determine best treatments) (Figure 2B)
  - The likelihood of knowing a lot/moderate amount was slightly higher in patients who were younger, Hispanic/Latino, or from an urban setting, or had a higher household income (Figure 3)
- 66% of patients' oncologists had talked to them about testing their tumor for mutations by a blood test or tumor biopsy
  - For more targeted mBC treatments, 60% said they would definitely prefer a blood test over tumor biopsy, while 28% would somewhat prefer a blood test and 11% did not have a preference

Awareness of *ESR1* mutations

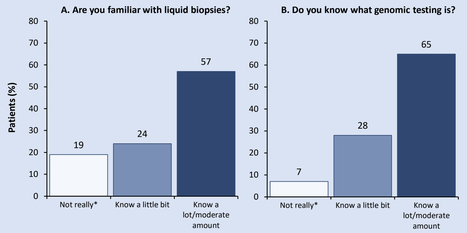
- Almost half of patients knew a fair amount about *ESR1* mutations and one-third a little bit (Figure 4)
  - Slightly more patients knew a lot/moderate amount about *ESR1* mutations (Figure 5) if they were 50 years or younger, Hispanic or Latino, or from an urban setting, or had a household income ≥$50K
- 45% of patients thought they had been tested for an *ESR1* mutation
  - More patients reported having undergone *ESR1* testing if they were younger vs older, Black vs Hispanic/Latino or White, or from an urban vs suburban setting (Figure 6)
- Most patients believed that *ESR1* testing results could affect their treatment options/decisions (95%), and were comfortable asking their providers about genomic and biomarker testing (96%)

Figure 1. First and second line treatment for mBC



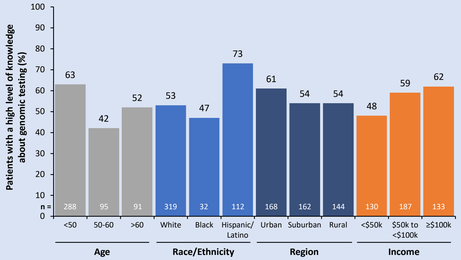
AI: anastrazole, letrozole, exemestane; SERM: tamoxifen; CDK4/6i: pablociclib, ribociclib, abemaciclib.
AI: aromatase inhibitor; SERM: selective estrogen receptor modulator; CDK4/6i: CDK4/6 inhibitor.

Figure 2. Knowledge of (A) liquid biopsies and (B) genomic testing in patients with mBC



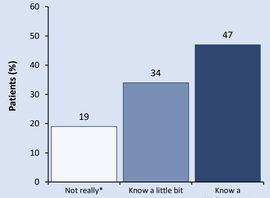
*Never heard of it or heard of it, but do not know what it is.

Figure 3. Patients with a high level of knowledge* about genomic testing stratified by demographic characteristics



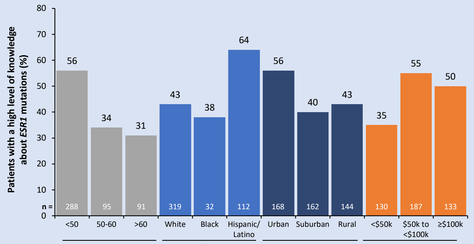
*Defined as knowing a lot/moderate amount of knowledge about genomic testing.

Figure 4. Do you know what an *ESR1* mutation is?



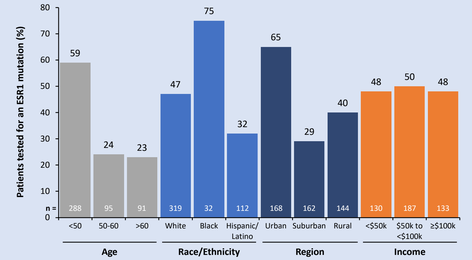
*Never heard of it or heard of it, but do not know what it is.

Figure 5. Patients with a high level of knowledge* about *ESR1* mutations stratified by demographic characteristics



*Defined as knowing a lot/moderate amount of knowledge about genomic testing.

Figure 6. Patients having been tested for an *ESR1* mutation stratified by demographic characteristics



Key Takeaways

- Awareness of genomic testing was high, but was lower for specific biomarkers such as *ESR1;* younger patients and Hispanic/Latino patients were more informed
- Patients are interested in liquid biopsies and would prefer them over tumor biopsies
- Achieving a diverse patient sample was made possible by reaching out to patient advocacy groups
  - This approach could be used to recruit for clinical trials and provide patient education

Conclusions

- In this survey of patients living with ER+/HER2- mBC, an unexpectedly high proportion of patients was familiar with liquid biopsies and genomic testing, but specific knowledge about *ESR1* mutations was slightly lower
- Concordance between physician discussion of genomic testing and liquid biopsies was observed
- *ESR1* mutation awareness analyzed by demographics suggests socioeconomic disparities in patient education and knowledge. Here, Hispanic/Latino versus White or Black patients and younger vs older patients were more likely to know about *ESR1* mutations
  - Knowledge about mutations in the Hispanic/Latino population was high, likely because most of these patients were recruited via FORCE, which is specifically for people with genetic mutations and hereditary cancer, hence increased awareness in this population
- Similar results were found for *ESR1* mutations testing, except more Black patients were tested than Hispanic/Latino patients
- Online surveys have several limitations, but our ability to reach this large (almost 500) ethnically diverse, mBC patient population over 4 months speaks to the value of our technique and the power of online mBC support groups
- This surveyed mBC population was relatively young, online-savvy with a high level of understanding of genomic testing, except for those from rural areas and with lower incomes
- Efforts to enhance education on genomic testing and *ESR1* mutations across different patient groups are needed, as genomic testing is becoming an important aspect of choosing mBC treatments

Disclosures

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