

Meta-Narrative Review of Possible Impacts of Genetic Screening on Treatment of Breast Cancer

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Introduction

According to Mahdavi & Nassiri, point mutations in genes like BRCA1 and BRCA2 have been established as being directly related to breast cancer in patients with a family history of breast cancer. Genetic screening of these genes is recommended if the patient has a family history of breast or ovarian cancer or if patient has been diagnosed with these cancers (National Cancer Institute, 2018).

There is no medical standard of prophylactic genetic screening for point mutation in breast cancer related genes in order to quantify a patient's potential risk of breast cancer development. According to the National Cancer Institute, the standard breast cancer preventative care methodology is to perform a mammogram every 1-2 years after age 40 and a yearly breast exam during the physical.

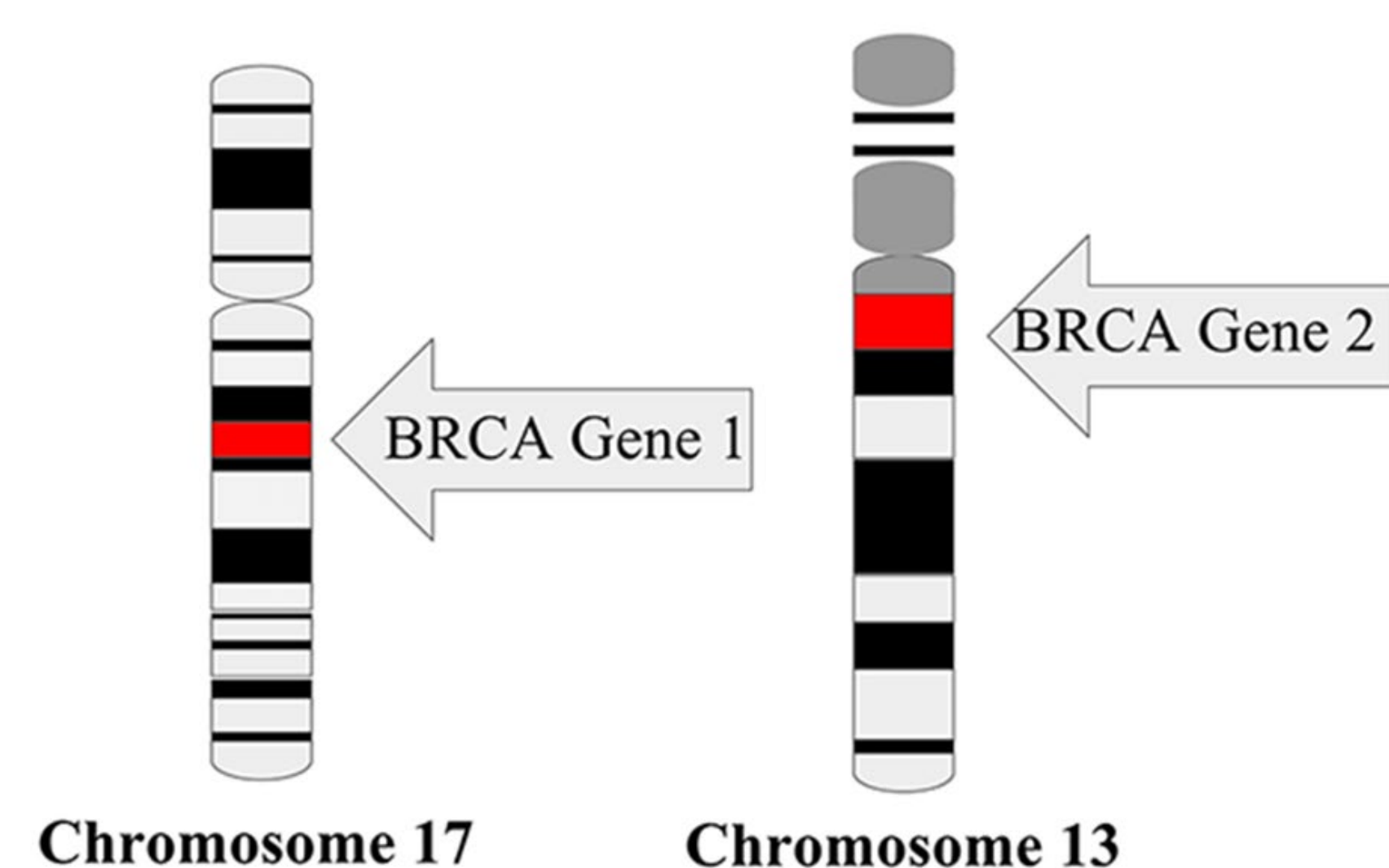


Figure 1. BRCA1/2 gene mutations on chromosomes (National Cancer Institute)

Current Screening Process for Breast Cancer

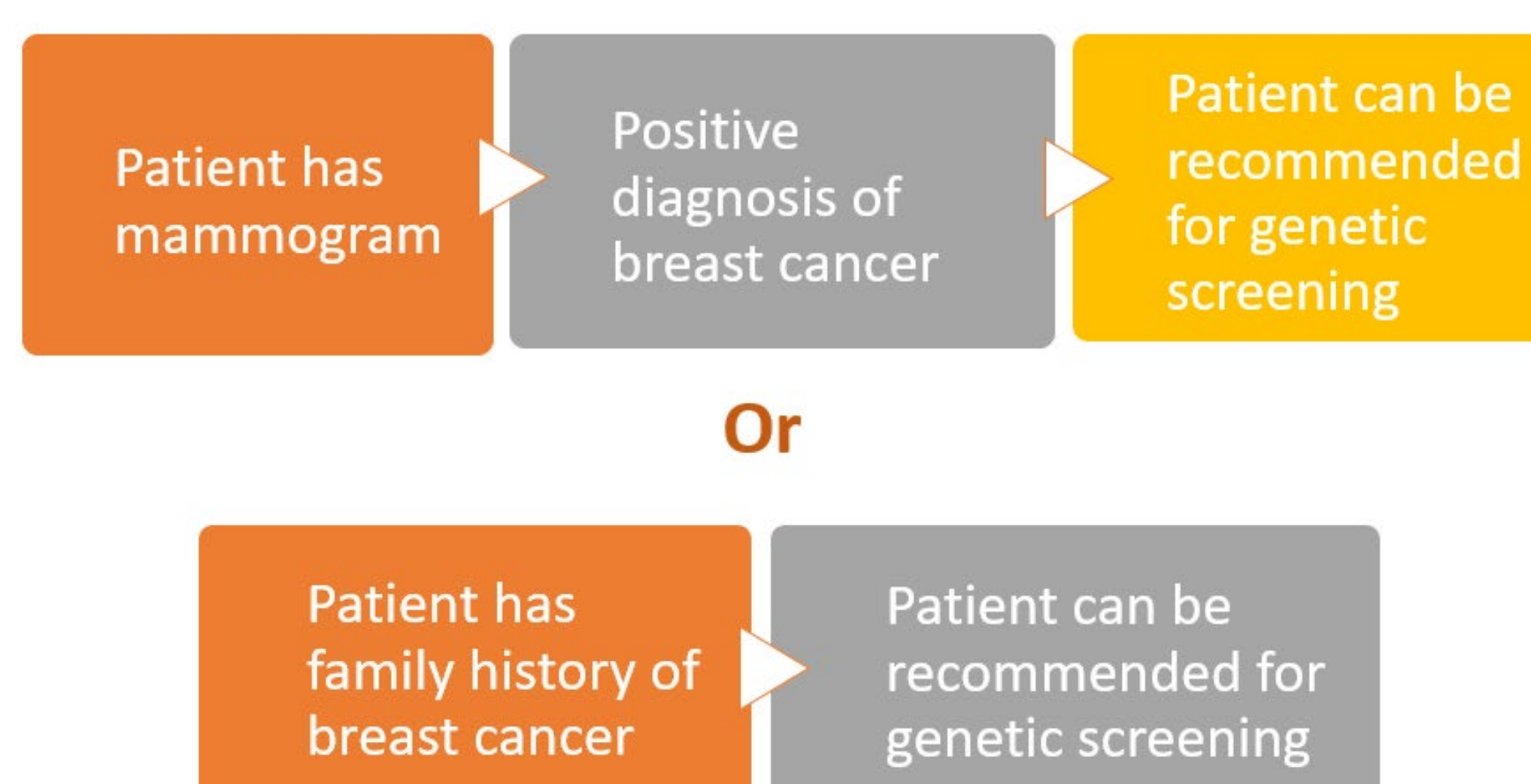


Figure 2. Visual of possible treatment pathways toward genetic screening in breast cancer according to Schwartz et al and the National Cancer Institute.

Research Question

Does genetic screening for breast cancer markers in patients with a family history of breast cancer affect patients' decisions in regard to treatment plans or surgical options?

Methodology

- Key Search Terms:
 - BRCA1/2, Genetic Screening, Breast Cancer, Genetic Markers, Barriers (with genetic screening), and Accessibility (with genetic screening).
- Search Publication Date: 2018 - Present
- Article Types:
 - Primary Research, Meta-Reviews, English Publication Language, Free Full Text, and Peer Review.
- Searches were not limited by country of publication nor country of the sample studied
- Article Narrowing Filters:
 - Pragmatic Clinical Trial, Clinical Study, Clinical Trial Phase 1-4, Observational Study, Multicenter Study, Case Reports, Classical Article, Evaluation Study, Research Support NIH Intramural, Research Support Non-US Govt, Research Support US Govt Non-PHS, Research Support US Govt PHS, Research Support US Govt, and Validation Study.
- Last Search Date: February 16, 2023.

Search Process

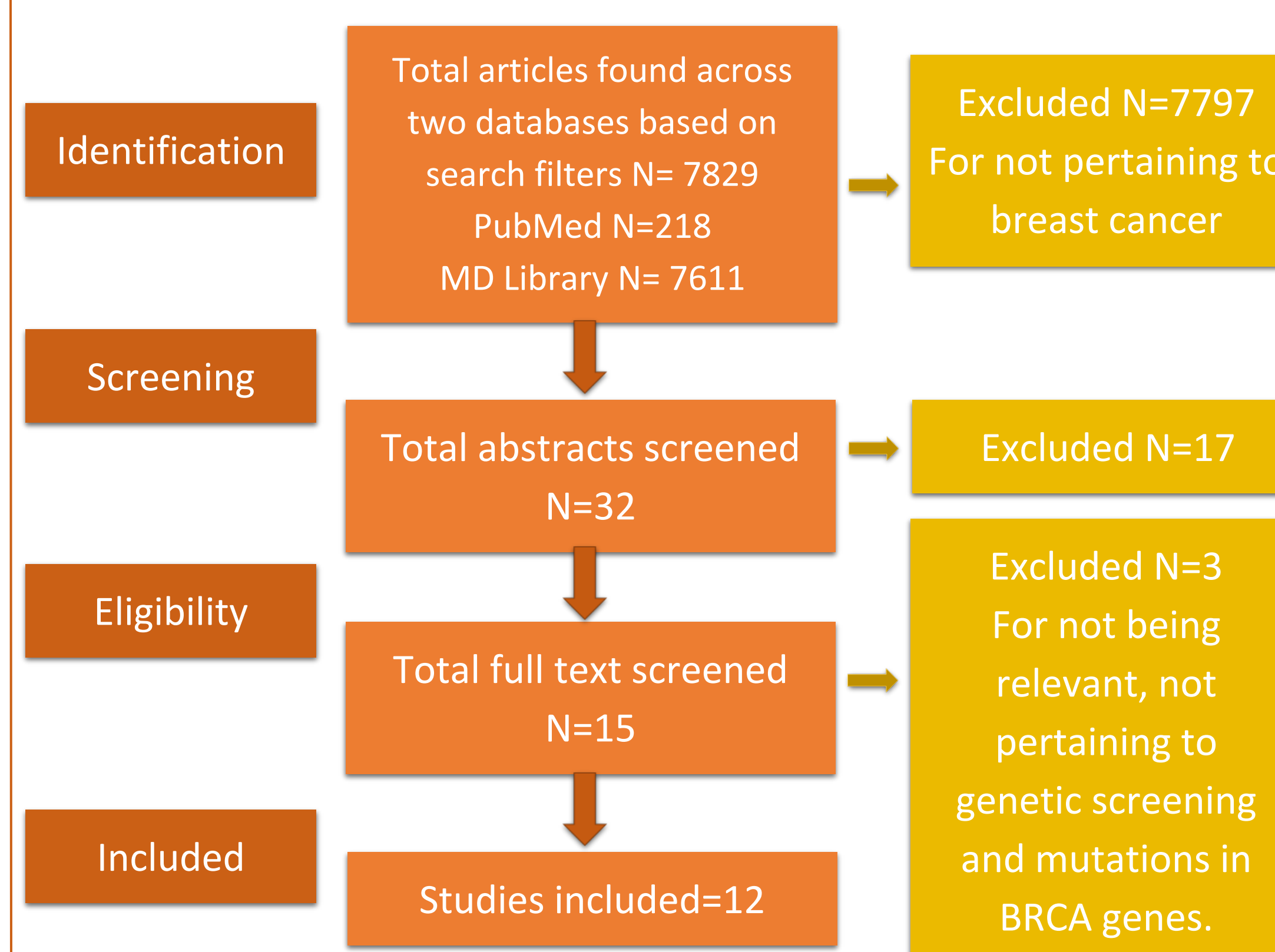


Figure 3. Assessment of document eligibility for meta-review research. This figure illustrates the guidelines for inclusion and exclusion at each review phase.

Results

Study Inconclusive

Current Process to Patient Genetic Screening has

Several Limiting Stages

- 1.Genetic Screening Not Recommended Until Patient Completes Genetic Counseling
- 2.Genetic Counselors are Scarce in Comparison to Labs Capable of Genetic Screening
- 3.Patient Must be Referred by Physician to Attend Genetic Counseling
- 4.Physician Must Have Compelling Evidence to Recommend Genetic Counseling
- 5.No Standard for Recommending Genetic Counseling, Case by Case Decision

Barriers to Genetic Screening Found in Multiple Articles

Suggested Racial Bias in Physician Referral for Genetic Counseling

Further Research Should Proceed Towards Validating Genetic Screening as Part of Standard Procedure in BRCA1/2 Mutation Related Breast Cancer Treatment

Figure 4. Visual representation of results using textboxes with high contrast colors. Genetic screening process according Schwartz et al.

Strengths and Limitations

Strengths:

- Shows increase in barriers to genetic counseling for groups that historically face healthcare disparities.
 - Awareness of barriers also increasing
- Demonstrates that progress is required but is not impossible for the American healthcare system.
- Shows that genetic counseling has positive psychological impact on patients (Nelson et al, 2019).

Limitations:

- Lack of robust research data associated with the research topic.
- Limited physician's recommendation rates
- Limited availability of genetic counseling.
- Impacts of genetic screening on BRCA-mutated breast cancer unknown due to lack of genetic screening.

Conclusion/Discussion

- Review inconclusive, insufficient literature
- Why is this research question difficult to answer?
 - Patients are unaware of genetic screening.
 - Physician bias (Ademuyiwa et al, 2021)
 - Socioeconomic status.
 - Social views on genetic screening.
- Limiting Stages Found in Review:
 - Genetic screening only recommended after genetic counseling (Schwartz et al, 2018)
 - Patient must be referred to genetic counseling by physician (Schwartz et al, 2018)
- Barriers Found in Review
 - Shortage of genetic counselors in comparison to labs capable of genetic screening limits patients in rural, low income, or low educational backgrounds from accessing genetic screening.
 - Racial bias of physicians referring their patients to genetic counseling, limits access to marginalized minorities that have higher rates of BRCA 1 or 2 mutated breast cancer.

Future Research

- Comparison studies of relapse rates and positive outcomes between patients that undergo genetic screening and those that do not.
- Research towards creating a universal standard procedure for broader inclusion of genetic counseling and screening in the breast cancer treatment pathway.
- Qualitative research identifying any biases or inequalities that can prevent patients from receiving genetic counseling or screening.
- Quantitative research for developing methods to reduce barriers to genetic screening that have been qualitatively identified.

References

- Ademuyiwa et al. Journal of Clinical Oncology 2021;39(36):4020-4028
 Nelson et al. JAMA 2019;322(7):666-685
 Schwartz et al. Breast Cancer Research and Treatment 2018;170(3):517-524
 National Cancer Institute. (2018). Breast cancer screening. Cancer.Gov; National Cancer Institute. <https://www.cancer.gov/types/breast/patient/breast-screening-pdq>