

Breast Cancer Detection and Prognosis using Mammographic and Genomic Data: A Comprehensive Review of Radiogenomics, Multimodal Deep Learning, and Explainable Artificial Intelligence

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Abstract— Breast cancer is a complex disease in that even its detection, molecular profiling, treatment selection and outcome prediction is challenging. Mammography can provide structure and morphology information to identify abnormal breast lesions, and genomic information can reveal molecular alterations, gene-expression profiles, tumour subtypes and disease stages. The integration of these complementary modalities has paved the way for innovative radiogenomics and multimodal AI solutions in BC. Here, we review recent progress in the use of mammographic and genomic data in tandem for detection of and prediction of breast cancer prognosis, focusing on radiomic feature extraction, genomic representation, multimodal data fusion, deep learning (DL), transformer-based architectures, and explainable artificial intelligence (XAI). Fusion strategies for incorporating imaging and genomic data on feature-level, decision-level, and attention-based levels are presented and evaluated. The review is on traditional datasets, evaluation metrics, data heterogeneity, missing modalities, small number of matched imaging-genomic cohorts, generalizability of the models and challenges interpreting the models. In addition, we discuss how explainable approaches, e.g. Grad-CAM and SHAP, can reveal clinically relevant imaging regions and genomic features. Lastly, promising avenues such as cross-modal learning, multi-omics data integration, self-supervised learning, domain adaptation, clinically interpretable multimodal models are discussed. – A review on the integration of mammographic and genomic data in a precision breast cancer framework is presented.

I. INTRODUCTION

Breast cancer is a biologically heterogeneous disease, and the diversity of tumor morphology, molecular characteristics, genetic aberrations, response to treatment, and clinical course hinders the ability to accurately predict the outcome for patients with this disease [1]. Mammography is an effective breast imaging technique to use for screening and diagnosis of breast cancer since breast tissue properties including tissue structure, masses, calcifications, density and other patterns in the tissue can be indirectly observed via X-ray. However, traditional mammographic evaluation is, in essence, the tumor's expressed phenotype, and may not accurately portray the tumor's molecular diversity. Recent advances in radiomics and artificial intelligence allow for the derivation of quantitative imaging features that may later be correlated with molecular and genomic data, offering new avenues for personalized breast cancer evaluation [2].

Genomic profiling also offers complementary insights by delineating gene-expression patterns, genetic mutations, molecular changes, and other biological processes linked to tumorigenesis. The combination of imaging phenotypes with genomic data is the foundation of radiogenomics aiming to correlate observable imaging phenotypes with the molecular biology of tumours [3]. Radiogenomic expression in breast cancer has more recently been explored for molecular subtype analysis, prognosis prediction, therapy response evaluation, and the detection of novel non-invasive biomarkers. While early studies focused predominantly on magnetic resonance imaging (MRI), radiogenomic studies have now been extended to include other breast imaging modalities such as mammography and digital breast tomosynthesis [4].

Deep learning [5] has broadened the scope for multimodal information integration in breast cancer studies. Convolutional neural network (CNN)-based, transformer-based, autoencoder-based, graph-based, attention-based and etc. algorithms are able to find complex medical data patterns. For example, a multimodal system could combine mammograms, genomic information, clinical data, pathology reports, and more. In general, a 3-level scheme is proposed by researchers to fuse these inputs, namely the feature level, decision level and the hybrid level. Still, this is an emerging area, partly since high-quality multimodal datasets are not, because data sources are heterogeneous, some types of information are missing, and models are challenging to validate outside of the context where they were developed [6].

This survey provides a cohesive perspective on the literatures pertaining to mammography, genomics, radiogenomics, multimodal deep learning, explainable ai. Instead of focus on imaging or genomics, this review republishes the transition from single modality-based analysis to multi-modality approaches, and points out the practical and methodological challenges in doing so. It includes feature extraction, data fusion, attention-based methodologies, and explanation tools (e.g., Grad-CAM, SHAP), as well as challenges pertaining to datasets, generalizability, and clinical use. A systematic review of the literature reveals that explainable multimodal methods are hindered by small or incomplete data sets, by inconsistent standards for evaluation, by missing data, and by limited validation in real world applications [7]. This review covers the application of mammograms and genomic profiles in each case to contribute to a more comprehensive characterization of breast cancer and to support clinical decisions. It also points out open research questions and possible future directions such as approaches that can deal with missing data modalities, incorporation of multi-omics data sources, adaptation to different clinical scenarios, models that directly capture uncertainty, privacy- preserving learning, and prospective clinical validation.

B. Objectives of the Review

The primary objective of this literature review paper is to:

- To summarize the state-of-the-art methods for detection and prediction of the progression of breast cancer from mammographic images. T
- To examine the relevance of genomic data and molecular characteristics in the diagnosis and prognosis of breast cancer.
- To explore the potential of radiogenomics and multimodal deep learning on the joint representation of mammographic and genomic data. To survey the application of explainable artificial intelligence in making mammographic and genomic predictions interpretable.
- To establish the key challenges and opportunities for future work in multimodal analysis of breast cancer.

Concept of Mammographic and Genomic Data

Mammographic and genomic results are two complementary sources of information for breast cancer investigation. Mammographic data describe the observable macrostructural and morphological features of breast tissue, whereas genomic data describe molecular and biological

features of tumour cells. The combination of these two data modalities is being developed as a cornerstone in radiogenomics and multimodal AI. Mammography is the most common method for breast cancer detection and screening, and genomic analysis can provide information for molecular classification, prognosis, therapy selection, and insights into tumor heterogeneity [8].

2.1 Concept of Mammographic Data

- Mammograms are a form of digital image using X-rays to examine the breast. This is a function of how much the tissues absorb the X-rays, which is characteristic of the type of tissue and in some cases can produce differences not visible to the naked eye. Radiologists search for density, masses, asymmetry, architectural distortion, and micro calcifications [9]. These are all attributes that determine the content of the description of the breast in the image, and can be used to spot suspicious regions that require attention.
- The state of the art in AI allows for the analysis of mammograms through various computational methodologies. Previously, computer-aided systems have been based on a limited number of predefined image features including texture, shape, intensity, edges, as well as statistical features. Advanced methods, e.g., Convolutional Neural Nets (CNNs) or transformer-based models, can learn multilevel features directly from images with more advanced representations. They help with visualization and segmentation of lesions, as well as with identifying regions of interest (ROIs) in a breast image, quantifying breast density, and assessing risk for breast cancer development [10].
- Mammograms reveal the appearance of breast tissue, but they don't explain everything. Tumours that look similar in an imaging test may have different molecular characteristics and may require different treatments. Conversely, tumours with similar molecular characteristics may look very different on a mammogram. This is a gap that scientists are trying to bridge by developing more accurate methods to correlate imaging data with genetic information [11].

2.2 Concept of Genomic Data

- Genomic data provide a description of the biological information stored within the genetic material of a patient's tumour cells. In breast cancer, genomics data can take the form of gene-expression (mRNA) profiles, DNA mutations, copy-number alterations,

DNA methylation, microRNA expression, and multiple other molecular features. This is important information for researchers to understand the biological processes of tumour formation, growth, metastasis, and response to therapy [12].

- Gene-expression profiles are so powerful because they represent the activity of many genes simultaneously. Differences in these signatures could potentially be used to differentiate breast cancer subtypes and to predict the behavior of a tumor. As such, genomic data may augment clinical and imaging findings by providing information on the biology of the disease [13].
- Yet, analysis of genomic datasets is not straightforward. They may have tens of thousands of genes but only a few samples of patients. Researchers use a variety of feature selection, dimensionality reduction, regularization, and deep learning methods to obtain the most relevant molecular features to address this imbalance and reduce overfitting.

2.3 Comparison of Mammographic and Genomic Data

Table 1. Comparison of Mammographic and Genomic Data in Breast Cancer Analysis

Characteristic	Mammographic Data	Genomic Data
Data type	Medical images	Molecular/biological data
Primary information	Structural and morphological characteristics	Genetic and molecular characteristics
Examples	Masses, calcifications, density, asymmetry	Gene expression, mutations, copy-number variation
Data representation	Pixels and image features	Genes, molecular variables and numerical expression values
Major analysis methods	CNN, Vision Transformer, radiomics	Statistical learning, autoencoders, DNN, transformers
Main applications	Detection, classification and	Subtyping, prognosis and molecular

	screening	characterization
Major challenge	Image quality, density and acquisition variation	Very high dimensionality and small sample size
Biological information	Indirect representation of tumour biology	Direct molecular information
AI integration	Image feature extraction	Genomic feature extraction
Combined potential	Provides phenotypic information	Provides molecular information

The table 1 highlights the complementarity between mammographic and genomic data. Mammographic data represents the breast tissue in anatomical and visual terms, whereas genomic data enables the representation of breast cancer in molecular and biological terms. Mammographic reading is primarily concerned with detection and diagnosis and genomic test results are employed to determine molecular profile and prognostic ate. Combining these two kinds of data could allow for a more complete representation of breast cancer, and it may enable breakthroughs in multimodal AI and radiogenomics.

Role of Multimodal Deep Learning and Explainable Artificial Intelligence

Multimodal deep learning and explainable artificial intelligence (XAI) are crucial for fusing and deciphering diverse data for breast cancer analysis. Mammographic data is visual and structural in nature, while genomic data is molecular representation of tumour features. Multimodal deep learning enables all these different types of data to be combined, while explainable artificial intelligence (XAI) allows scientists and clinicians to understand the factors driving model predictions [14].

3.1 Role of Multimodal Deep Learning

Multimodal deep learning is a specific type of artificial intelligence method, which can simultaneously model two or more types of data. For breast cancer, mammogram images and genomics data can be analyzed using different learning networks and then combined into a unified representation. Different types of AI models can be used to analyze mammograms and genomic data. Researchers can use convolutional neural networks or vision transformers

for images, and deep neural networks, autoencoders, or transformers for genomic data [15]. The resulting representations can be integrated through feature combination, decision-level fusion or attention-based techniques. Combining the two sources can expose information that would not be detected with either source alone. Mammograms detect physical changes, like lumps or calcium deposits, while genomic data may provide insights into a tumour's molecular profile and behaviour. Therefore, their combination may lead to better breast cancer classification, molecular subtype prediction, recurrence prediction and prognosis [16].

Multimodal learning can also overcome the limitations of individual data sources. An example might be an imaging model that identifies a suspicious lesion but provides limited information about its molecular characteristics. Conversely, genomic analysis can identify molecular changes but provides no information about the spatial appearance of the tumour. This fusion of modalities presents a chance to build a more complete representation of the disease.

3.2 Role of Explainable Artificial Intelligence

Explainable artificial intelligence (XAI) refers to methods and techniques in the application of AI such that the results of the solution can be understood by human experts. The need for explainability is particularly critical in medical applications where model outputs must be understandable to, and be critically appraised by, clinicians prior to being used in clinical decision making [19].

In mammographic analysis, Grad-CAM and similar methods can be applied to show which parts of the image contributed the most to the prediction of a model. This can show if the model is learning to predict on something informative such as a suspicious mass or calcification or any other area. In genomic applications, methods like SHAP can identify genes or molecular features that have a significant impact on a prediction. Explainability can be introduced to the multimodal learning, where the model can give explanations in two modalities. As an illustration, the system might identify a region in a mammogram as being significant, and point to a group of genomic features that are associated with the predicted risk. This may contribute to enhancing transparency and, enabling better interpretability even for complicated multimodal models [20].

Table 2. Role of Multimodal Deep Learning and Explainable Artificial Intelligence in Breast Cancer Analysis

Technology	Role	Possible Application
Multimodal deep learning	Integrates different data modalities	Mammogram–genomic analysis
Convolutional neural network	Extracts image features	Lesion detection
Vision Transformer	Learns global image representations	Mammographic classification
Deep neural network	Processes genomic variables	Molecular prediction
Attention mechanism	Identifies important cross-modal information	Multimodal fusion
Grad-CAM	Highlights important image regions	Mammogram interpretation
SHAP	Identifies influential features	Genomic interpretation
Multimodal XAI	Explains information from multiple modalities	Clinical decision support

The table 2 indicates that multimodal deep learning and explainable artificial intelligence serve different yet complementary roles. Multimodal learning is about fusing and learning on mammographic and genomic data, while explainable artificial intelligence is about the understanding the rationale behind the resulting predictions. Coupled, these technologies could enable a more window-like, interpretable, and holistic breast cancer analysis.

II. METHODOLOGY

This review article provides a systematic review of advances in breast cancer detection and prognosis based on mammographic and genomic information, focusing on radiogenomics, multimodal deep learning, and interpretable AI. The approach was intended to capture mammographic and genomic analysis research that is the most relevant and high quality, as well as to consider the progression of such research and the evaluation of methods based on imaging

and molecular data. A process of systematic literature preselection was performed to find up-to-date research and to keep a few previously selected seminal work for theoretical and methodological background.

A. Databases/Resources

Research for this review was drawn from major academic databases and scientific publishing platforms covering medical imaging, cancer research, genomics, and artificial intelligence. Sources included:

- IEEE Xplore
- SpringerLink
- ScienceDirect
- Wiley Online Library
- PubMed
- ACM Digital Library
- Google Scholar

These sources were chosen for their broad coverage of work on breast cancer, mammography, genomic analysis, radiomics, radiogenomics, deep learning, multimodal AI, and explainable AI. Relevant studies were also found by checking the reference lists of review articles and influential research papers, as well as by following their citations.

B. Inclusion Criteria

Rigorous inclusion criteria were implemented to guarantee the relevance of the studies selected for the review with respect to its objectives. The selection criteria were formulated to exclude studies lacking the necessary evidence strength or key deliverables. It was decided to concentrate on the period from 2020 to 2026 due to the fluidity of the field and the need to capture the latest advances in AI, radiogenomics, multimodal learning, and xai; however, earlier studies were memorized when they were indicative of fundamental processes related to mammography, rad iomics, genomics, or radiogenomics. The studies had to concern breast cancer detection, classification, molecular characterization, prognosis, prediction of recurrence, risk assessment, or prediction of response treatment. Studies concerning mammography, digital mammography and mammographic images or quantitative imaging features that were included in the studies were determined to be applicable. Investigations of genomics and molecular data, such as gene expression, genetic alterations, molecular subtypes, copy-number variants, transcriptomic data, and other genomic features, were also eligible for inclusion. In addition, studies that use

AI and ML based methods such as DL, convolutional neural network (CNN), vision transformer (ViT), autoencoders (AE), attention mechanism (AM), graph neural network (GNN), and multimodal solution were included. Special emphasis was placed on research using xai methods, Grad-CAM, SHAP, LIME and attention-based visualizations, to enhance the interpretability of predictions of breast cancer. Studies that were relevant exclusively to other cancers, non-medical applications, or breast cancer applications that did not meaningfully involve mammographic data, genomic data, radiogenomics, multimodal learning or xai were excluded.

C. Search Keywords

A selective keyword method was applied to obtain pertinent literature. The main keywords and phrases were:

- Detection of breast cancer
- Prognosis of breast cancer
- Breast cancer and mammography
- Breast cancer screening: review of protocols in the context of radiological and mammographic image analysis
- Breast cancer and genomic data
- Genomics of breast cancer
- Radiomics in breast cancer
- Radiogenomics in breast cancer
- Mammography through genomics
- Multi-modal deep learning
- Breast Cancer - Multimodal Analysis (B-CAMA)
- Explainable AI for breast cancer
- XDL: Explainable deep learning for medical diagnosis
- Artificial intelligence and radiogenomics
- Genomics of imaging
- Multi-omics breast cancer

Using Boolean operators such as AND, OR spinners to formulate refined search queries. Some of the related searches were “breast cancer” AND “mammography” AND “deep learning”, “breast cancer” AND “genomic data” AND “radiogenomics”, and “mammography” AND “genomics” AND “multimodal artificial intelligence”.

D. Step-wise Selection of Literature

- A step-wise literature selection process was used to select the studies that were most pertinent to the aims of the review.
- **Data Collection:** The search process for the selected databases was comprised of multiple phases beginning

with the definition of key terms and sample phrase combinations. We acquired additional potentially relevant studies by citation tracking and reference-list screening. The first search returned on the line about 100 articles.

- **Screening:** Titles and abstracts were screened against predefined inclusion and exclusion criteria in the second phase. Articles were included if they pertained to breast cancer, mammographic data, genomic data, radiogenomics, multimodal learning or XAI. As a result of this screening, 25 articles were shortlisted.
- **Final Review:** Subsequently, full texts of the selected studies were reviewed more thoroughly. Studies that have made substantial contributions to mammographic analysis, genomic profiling, radiogenomics, multimodal dL, and XAI were given special attention. A total of 15 studies were chosen for comprehensive evaluation in the final review after this appraisal, as in Table 3.

Table 3. Selection Process of Literature Review

Stage	Number of Papers	Description
Initial Collection	100	Papers identified through database searches, keyword combinations, reference screening, and citation tracking related to mammography, genomics, radiogenomics, multimodal deep learning, and explainable artificial intelligence.
Shortlisting	25	Papers screened according to relevance to breast cancer, mammographic analysis, genomic data, artificial intelligence, radiogenomics, and multimodal approaches.
Final Review	15	Studies selected for detailed analysis based on methodological relevance, contribution, data characteristics, artificial intelligence techniques, evaluation methods, and significance to the objectives of the review.

Table 3 shows the systematic reduction of literature from the initial collection to the final set of studies examined in detail. Indeed, a large number of potentially relevant articles was found via the initial search, articles were omitted for one or more of the following reasons: Based on the title and abstract, were not related to the aim of the review or they did not meet the predefined inclusion criteria. The criteria for final selection of inclusions were the relevance of the study focus on the fusion of mammographic and genomic data, radiogenomics, multimodal deep learning, and interpretable artificial intelligence. This screening procedure provided for a focused coverage of reviews on novelty and key studies.



Fig.1. Flowchart for Systematic Review

The selection process can be reduced to a three-stage funnel on the basis of that initial 100: Screening (first cut with 30 papers) → Review all the papers (25) → last review (15). The narrowing funnel, if you will, is the manner in which relevance, quality, methodology and thematic criteria were progressively imposed to sift out the studies that best suited the purposes of this review article.

III. LITERATURE REVIEW

The final selected papers for literature review are as follows,

Table 3. Summary of Studies

Author (Year)	Summary of Work
Sabit et al. (2026) [21]	Reviewed the integration of multi-omics and AI for personalized breast cancer management, including genomics, transcriptomics, proteomics, and metabolomics for diagnosis, prognosis, and treatment prediction.
Jiang et al.	Reviewed AI and multi-omics convergence

(2026) [22]	in breast cancer, emphasizing multimodal integration of molecular, imaging, pathology, and clinical information for diagnosis and prognosis.
Kumari et al. (2026) [23]	Reviewed AI-driven multi-omics applications for breast cancer screening, diagnosis, patient stratification, biomarker discovery, and therapeutic-response prediction.
Pesapane et al. (2026) [24]	Systematically reviewed explainable AI in mammography and found that XAI methods require stronger external validation and clinical evaluation despite promising visual explanations.
Li et al. (2026) [25]	Developed a multimodal deep-learning system for non-invasive breast cancer diagnosis using mammography, ultrasound, MRI, and radiology-pathology alignment.
Hezil et al. (2026) [26]	Proposed selective multimodal deep learning for breast cancer subtype classification by combining histopathological and genomic information.
Sucre et al. (2025) [27]	Compared multimodal fusion strategies for breast cancer survival prediction using multi-omics data and showed the importance of appropriate fusion strategies and explainability.
Li et al. (2025) [28]	Reviewed deep learning approaches for multimodal breast cancer data fusion and classified approaches into feature-level, decision-level, and hybrid fusion strategies.
Shifa et al. (2025) [29]	Reviewed explainable AI techniques for mammographic breast cancer screening and emphasized the need for standardized evaluation of model explanations.
Kandan et al. (2025) [30]	Developed a multi-input CNN approach for breast cancer prognosis prediction using multimodal information, demonstrating the potential of combining different data sources for outcome prediction.
You et al. (2024) [31]	Developed a multicenter radio-multiomic approach for breast cancer outcome prediction and investigated relationships between imaging characteristics and biological information.

McAuliffe et al. (2025) [32]	Conducted a systematic review and meta-analysis of radiogenomic profiling for determining BRCA alteration status, demonstrating the potential of imaging-derived characteristics for non-invasive genomic assessment.
Donnelly et al. (2024) [33]	Developed AsymMirai, an interpretable mammography-based deep-learning model for predicting one-to-five-year breast cancer risk using bilateral mammographic differences.
Balve and Hendrix (2024) [34]	Compared LIME, Grad-CAM, and Kernel SHAP for interpreting CNN-based mammographic breast cancer classification, with Grad-CAM providing useful visualization of model behaviour.
Wang (2024) [35]	Reviewed deep-learning applications in mammography for breast cancer detection and classification, highlighting opportunities in risk prediction, prognosis, interpretability, and clinical implementation.

The studies under review in Table 3, clearly indicate the transition from classical, single-modality analysis of breast cancer to multimodal, integrated AI solutions. Recent studies have suggested that mammographic images contain information about tumour morphology and breast tissue patterns, whereas genomic and multi-omics information contain data related to molecular behaviour, subtype, prognosis, and treatment response. Results You et al“ (2024)McAuliffe et al“(2025)Li et al“(2025)Empirical studies also indicate the increasing relevance of bridging imaging phenotypes with molecular data via radiogenomic and multimodal methodologies. Likewise, the work of Sucre et al. (2025) and Hezilet al.(2026) predicts the promise of multimodal fusion in prognostic, and molecular subtype based, classification. Explainability based AI has gained more attention with works of Balve and Hendrix , Shifa et al and Pesapane et al. emphasizing the need to ability to understand model decision as opposed to relying on predictive performance. Nevertheless, several challenges, such as the scarcity of matched imaging–genomic data sets, the high dimensionality of genomics, the presence of missing modalities, differences among data sets, the lack of rigorous external validation, and limited clinical evaluation of methods to explainability, are identified in literature and

remain persisting problems. The available work thus testifies the feasibility of the mammographic and genomic feature fusion, yet a unified framework between radiogenomics, multi-modal deep learning and explainable AI remains a promising yet challenging research direction.

IV. CONCLUSION

This review discussed the emerging trend of data integration of mammographic and genomic data in breast cancer detection and prognosis, mainly in view of radiogenomics, multimodal deep learning, and explainable artificial intelligence. The cited studies support that mammographic data is informative of the structural and morphological properties of breast tissue while genomic data can reveal complementary information on molecular features, tumour heterogeneity, disease progression and prognosis. Probably, multimodal deep-learning methods can leverage these complementary information at the feature-level, decision-level, and attention-based fusion level, facilitating ageneral modelling of breast cancer which is not achievable by single modality. Meanwhile, explainable artificial intelligence methods, e.g., Grad-CAM and SHAP, can enhance the interpretability of model predictions by highlighting the contributing imaging regions and genomic features. Despite these advances, there remain significant challenges, such as scarce matched mammographic–genomic data sets, high-dimensional genomic data, absent modalities, data heterogeneity, limited external validation, and concerns on the clinical reliability of explanations. Therefore, the future investigation should be directed towards strong multimodal architectures, big data and data diversity, cross-modal learning, multi-omics data integration, missing data imputation, domain adaptation, appropriate evaluation which is standardized, and clinically validated explainability. As such, the inclusion of mammographic and genomic data appears to be a promising avenue for achieving more comprehensive, explainable and individualized Artificial Intelligence (AI)-based breast cancer detection and prognosis.

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