Review

Journal of INTERNATIONAL MEDICAL RESEARCH

Journal of International Medical Research
2024, Vol. 52(4) 1–29

DOI: 10.1177/03000605241237867
journals.sagepub.com/home/imr
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Machine learning and new insights for breast cancer diagnosis

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Abstract

Breast cancer (BC) is the most prominent form of cancer among females all over the world. The current methods of BC detection include X-ray mammography, ultrasound, computed tomography, magnetic resonance imaging, positron emission tomography and breast thermographic techniques. More recently, machine learning (ML) tools have been increasingly employed in diagnostic medicine for its high efficiency in detection and intervention. The subsequent imaging features and mathematical analyses can then be used to generate ML models, which stratify, differentiate and detect benign and malignant breast lesions. Given its marked advantages, radiomics is a frequently used tool in recent research and clinics. Artificial neural networks and deep learning (DL) are novel forms of ML that evaluate data using computer simulation of the human brain. DL directly processes unstructured information, such as images, sounds and language, and performs precise clinical image stratification, medical record analyses and tumour diagnosis. Herein, this review thoroughly summarizes prior investigations on the application of medical images for the detection and intervention of BC using radiomics, namely DL and ML. The aim was to provide guidance to scientists regarding the use of artificial intelligence and ML in research and the clinic.

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Keywords
Breast cancer, machine learning, radiomics, artificial intelligence, deep learning

Date received: 21 August 2023; accepted: 21 February 2024

Introduction

Breast cancer (BC) is the most prominent malignant tumour among females worldwide and it accounts of almost 10.4% of all cancers.[1] It is characterized by an aberrant, disorderly, and invasive proliferation of breast cells. Moreover, BC cells can readily escape to the circulatory or lymphatic system, where they generate new tumours and invade distant vital organs.[2] At present, BC is the largest contributor to cancer-related mortality among women between 20–50 years of age. Based on information from the American Cancer Society, in 2019 alone, there were an estimated 268 600 new incidences and 41 740 cancer-associated deaths in the United States.[2] Alarmingly, in 2020, the cancer-related mortality among women increased to 684 996 global deaths, making it the major contributor of deaths across the globe.[1] In addition, in 2021, the World Health Organization announced BC as the most prevalent form of global cancer, far exceeding lung cancer.[1] At present, these numbers are rapidly increasing, and a 50% rise is predicted over the next two decades owing to enhanced life expectancy, unhealthy diets, inadequate physical activity and detrimental substance intake (for example, alcohol).[1] Given these critical factors, there is an urgent need for extensive research in all areas of BC, from prevention to early detection and efficacious intervention.[2,3]

Personalized BC interventions are highly dependent on accurate diagnosis. BC typically features discrete histological, molecular and clinical phenotypes; and it sometimes manifests with radiological heterogeneity. Nevertheless, classical detection techniques do not provide adequate information for proper BC diagnosis.[4] Radiomics is a relatively new tool for the extensive analysis of medical images. The subsequent imaging features and mathematical analyses can then be used to generate machine learning (ML) models, which stratify, differentiate, and detect benign (b-BT) and malignant breast lesions (m-BT), which, in turn, can be used to establish risk and formulate the optimal intervention for patients with b-BT and/or m-BT. Relative to classical influential science and skilled radiologists, radiomics features offer a more precise diagnostic method of BT detection. Hence, radiomics-based explorations/research are well underway in different parts of the world.[5]

More recently, scientists increased the application of ML tools, namely, image segmentation, stratification and estimation for BC assessment. Image segmentation both identifies and extracts tumour and its adjoining normal tissue. ML tools, particularly, convolutional neural networks (CNNs), are widely employed for automated BC segmentation. For example, a previous study established a CNN-based algorithm, with a mean dice similarity coefficient of 0.85, to segment BC tumours in ultrasound images.[6] Stratification delineates between m-BT and b-BT and predicts the probability of recurrence or metastasis. ML tools, namely, support vector machines (SVMs) and random forests (RFs), are frequently applied in BC stratification. For example, previous research established an RF-derived algorithm for breast tumour stratification of mammograph images, using a sensitivity (SEN) and specificity (SPC) of 87.5% and 91.7%, respectively.[7] Prediction estimates the probability of tumour recurrence or metastasis, as well as treatment response. ML techniques, namely, deep neural networks (DNNs) and recurrent neural networks (RNNs), are also widely employed for BC estimation. For example, a study reported a DNN-derived method for estimating BC recurrence risk, with an area under the receiver operating characteristic curve (AUC) of 0.87.[8]

Medical imaging, namely, X-ray mammography (MG), magnetic resonance imaging (MRI) and positron emission tomography (PET), offers a non-invasive approach of extracting data on BC morphology, metabolism and blood flow. Radiomics, a progressive tool for the isolation of quantitative profile from medical images, has garnered much attention owing to its ability to retrieve both spatial heterogeneity and functional profiles of tumours. BC imaging has made remarkable recent progress, with marked advancements in both technology and imaging analyses. The common approaches of BC detection encompass MG, ultrasound (US), MRI and molecular imaging. Using these techniques, it is now possible to detect and diagnose early-stage BC, which ultimately enhanced patient prognoses. Moreover, ML and deep learning (DL) algorithms are also applied in BC image analysis, with promising results. Herein, this review presents a detailed summary of the current ML applications for BC imaging, namely, MG, ultrasound, MRI, PET and others. The article selection process is detailed in supplemental information. Based on the module and search string in PubMed® (see supplementary materials, Table S1) and Web of Science™ (see supplementary materials, Table S2), a retrieval strategy of the above respective databases was established (see supplementary materials, Table S1). Overall, 158 articles were selected from the date of inception of the databases up to 31 May 2023. Figure 1 shows the results of the literature search and selection.

X-ray mammography

X-ray mammography is routinely used to screen and diagnose BC, and it is a well-established method of minimizing cancer-associated deaths.[9] BC tumours are initially screened using X-ray mammography, prior to manual interpretation by a radiologist to estimate whether it is b-BT and m-BT. The popularity of MG stems from its reduced-dose X-ray, enhanced-contrast, augmented-resolution detectors and X-ray system particularly intended for breast imaging. In general, MG can be separated into two groups: film screen MG (FSM), whereby a film screen serves as the end-recording device; and full-field digital MG (FFDM), whereby digital detectors serve as the recording media. Digital images from FFDM provide numerous benefits over FSM, particularly in terms of ease of image processing and enhancement.[4]

In recent times, there has been a considerable rise in artificial intelligence (AI) algorithm use in medicine. The available AI includes radiomics and DL, which markedly enhances lesion detection and diagnosis from medical images in clinical practice.[10,11] The current AI is rather sophisticated and approaches the performance of a skilled radiologist, particularly in MG.[12] MG images, photographed and quantified using radiomics, offer a remarkable diagnostic capacity for b-BT and m-BT, and they also provide other relevant data to radiologists.[4] To further augment MG screening accuracy, the computer-aided diagnosis system (CAD) software 4 was designed for BC tumour identification and used since the 1990s. Regrettably, information from earlier CAD systems did not yield any marked improvement in patient outcome.[13,14] With significant advancements

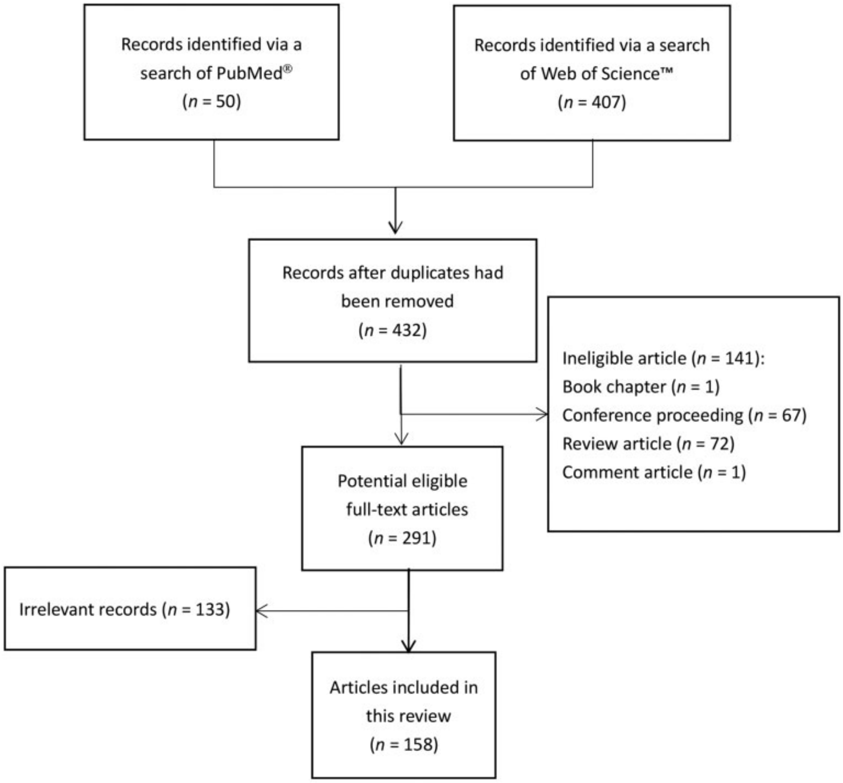

Figure 1. Flow diagram showing the identification of the research articles describing the current machine learning applications for breast cancer imaging that were included in this review.

in DL visual object identification, and multiple other domains, more scientists are showing interest in establishing DL tools to augment the MG screening of women.[15–18] Emerging evidence suggested that the performance of a DL-based CAD system was comparable to the radiologist performance alone, and enhanced the radiologists' performance in a supporting mode.[19,20] One study introduced a collaborative DL method that clearly classified pathological images into cancerous and non-cancerous BC tissues on 544 complete whole slide images, with SEN and accuracy of 97.73% and 95.29%, respectively.[19]

Machine learning is a form of AI introduced in the 1980s. ML primarily examines how computers mimic human learning behaviours, procure novel information, enhance existing information, as well as their own performance. ML conducts tasks without clear programming directions, namely, it identifies hidden associations between data and analyses them. Among the common ML applications are logistic regression (LR), linear regression, decision trees, RFs, naive Bayes and K-means cluster analyses, multilayer perceptron (MLP) and SVMs.[21] Artificial neural networks (ANN) and DL are novel areas of ML and these tools analyse data using computer simulation of the human brain. ANN is based out of the biological learning mode of human brain neurons that interconnect with one another, and regulate cascade, alteration and classification. DL is more advanced than ANN. DN utilizes the hierarchical ANN to establish highly complicated learning models that elucidate data in various dimensions. DL encompasses a dynamic Bayesian network, CNN and RNN. The CNN algorithm distinctly benefits image processing and is employed in feature isolation and analysis of clinical imaging information. Recursive neural network algorithms dynamically monitor disease via time series data analysis. Traditional ML requires feature extraction from the original data, as well as processing into structured datasets, which cannot directly process unstructured data. DL also directly processes unstructured information, such as images, sounds and language; and is highly beneficial in clinical image stratification, medical record analysis and tumour diagnosis.[22,23] Thus, multiple strategies are implemented in mammogram diagnosing. These can be classified into statistical-, wavelets-, Markovian- and machine-learning-based methods.[24] A previous study developed a region of interest (ROI)-based CNN termed You Only Look Once, which simultaneously identified and categorized breast masses into digital mammograms.[25] This model was composed of distinct phases, namely, preprocessing, feature retrieval, mass detection with belief and tumour stratification with fully connected neural networks. However, this model was not appropriate for a small dataset and the tumour area was not partitioned within mammograms. To classify MG mass lesions and examine the CNN model, researchers developed deep convolutional neural networks.[21] Upon preprocessing and normalization of isolated ROIs from the entire mammogram, the ROIs were integrated to form a unified dataset, which was then used to update the CNN. However, this technique did not minimize noise or muscles, which potentially resulted in mis-stratification.

Machine leaning, particularly breast imaging CAD system, aids in BC tumour diagnosis, and it is not influenced by the radiologist's reading mode, fatigue, distraction and other factors. Therefore, this system greatly enhances SEN of BC diagnosis.[22] Researchers examined an AI system's aptitude to substitute for doctors in BC diagnosis.[19] The study revealed that the AI systems are quite efficient, relative to radiologists, in BC detection.[19] Another study also evaluated cytological image analysis for BC identification and stratification using Naive Bayesian and ANN, demonstrating 98% precision in BC detection.[23] Similarly, Another study reported an enhanced crow search optimized extreme learning machine process, with approximately 98.26%, 97.193% and 98.137% accuracies for the reast, DDSM and MI - AS datasets, respectively.[26] Researchers developed a CAD system involving two components as follows: the first component identified an ROI; and the second extracted relevant profiles utilizing CNN.[27] Finally, using support vector machine, 87.2% accuracy was achieved in BC estimation from mammograms.[27] Another study also presented a CAD system for BC detection from MG images.[24] Their method manipulated MG images for ROI identification, relevant profile extraction and optimization, and BC stratification using a specific ML algorithm, SVM.[24] Based on their results, the aforementioned approach exhibited strong efficacy, particularly in distinguishing between normal and abnormal tumours with 100% accuracy.[24] Lastly, researchers employed an integrated regression learning, SVM and MLP system to stratify mammograms.[28] They yielded 99.42% accuracy with the Wisconsin Breast Cancer Dataset.[28] This current review extracted and organized the data in tabular form and summarized the use of MG in the diagnosis of breast cancer (Table 1).[2,4,5,7,9,24,29–79]

Table 1. Models, classes and performance for breast X-ray mammography data in selected papers.[2,4,5,7,9,24,29–79]

Paper reference	Models/algorithm	Binary or multiclass	Classes	Accuracy	Other performance evaluation parameters	Anomaly application/task
29	LIBSVM	Binary	Positive/negative	–	AUC = 0.725 ± 0.018	BC risk detection
30	SVM	Binary	Malignant/benign	99.385%	Sensitivity = 100% Specificity = 98.785%	BC detection
31	SVM, ELM	Multiclass	Normal/benign/malignant	94.11%	–	BC detection
32	DL, SAE	Binary	Malignant/benign	89.7%	AUC = 0.90	BC detection
33	dANN	Binary	Malignant/benign	0.81	AUC = 0.85	BC detection
34	DCNN	Binary	Malignant/benign	0.85	AUC = 0.84	BC detection
35	DNN	Binary	Malignant/benign	–	–	BC tumour detection
36	SVM, LASSO, KNN, B:SWIMS	Binary	Malignant/benign	0.738	Sensitivity = 0.957	BC tumour detection
37	SVM	Binary	Malignant/benign	0.90	ROC = 0.95	BC tumour detection
38	SVM, KNN, LDA	Binary	Malignant/benign	0.80	Specificity = 0.94 ± 0.06 Sensitivity = 0.66 ± 0.24	BC diagnosis
7	RF	Binary	Malignant/benign	0.93	Sensitivity = 0.91 Accuracy = 0.82	BC tumour detection
4	SVM, KNN, LR	Binary	Malignant/benign	0.978, 0.975	Specificity = 0.975 Sensitivity = 0.983	BC diagnosis
39	RF	Binary	Normal/tumour	0.61	Sensitivity = 70% Specificity = 49%	BC risk outcome
40	CNN, VGG16, residual network	Binary	Normal/cancer	0.98	Specificity = 0.91 Sensitivity = 0.87	BC tumour detection
41	Faster R-CNN	Binary	Lesion/non-lesion Malignant/benign	–	–	BC tumour detection
42	SVM, VGG16, CNN	Binary	Malignant/benign	0.88	AUC = 0.88	BC image classification
5	SVM, RF, NB	Binary	Malignant/benign	0.72	AUC = 0.79	BC image classification
43	DCNN	Binary	Malignant/benign	–	–	BC image classification BC diagnosis
44	Faster R-CNN30, CNN	Binary	Malignant/benign	0.94	Specificity 0.92	Cancer subtype classification
45	DNN	Binary	Malignant/benign	0.92	AUC = 0.92	BC image classification
46	ANN	Binary	Malignant/benign	0.82	AUC = 0.83	Identify risk categories

(continued)

Table 1. Continued.

Paper reference	Models/algorithm	Binary or multiclass	Classes	Accuracy	Other performance evaluation parameters	Anomaly application/task
47	VGG16, SVM, KNN, NB, DT	Binary	Malignant/benign	0.99	Specificity = 0.98	BC tumour detection
48	SVM	Binary	Malignant/benign	0.82	AUC = 0.80	BC tumour detection
2	DCNN, VGG19	Binary	Malignant/benign	0.99	Specificity = 1.00	BC image classification
49	DCNN	Binary	Malignant/benign	0.80	ROC 0.84	BC tumour detection
50	RF	Multiclass	Normal/benign/malignant	0.85	F-score 0.98	BC tumour detection and classification
51	SVM, QGA	–	–	–	–	BC tumour edge detection
52	DNN, AlexNet, VGGNet	Binary	Malignant/benign	0.987	Specificity = 0.99 Sensitivity = 0.99	BC classification
53	RBFNN	Binary	Malignant/benign	0.98	Sensitivity = 0.98	Breast microcalcifications early diagnosing
54	DCNN	Binary	Malignant/benign	–	AUC = 0.91	Breast mass classification
55	ANN	Binary	Malignant/benign	–	Specificity = 0.94	BC screening and diagnosis
56	ANN, SVM	Binary	Malignant/benign	0.99	Specificity = 0.99 Sensitivity = 0.99	BC tumour detection
24	SRG, SVM	Binary	Malignant/benign	0.87	Specificity = 0.86 Sensitivity = 0.90	BC tumour detection
57	SVM, NSEA	Multiclass	BI-RADS classification	0.94	Time = 0.68 s	BC diagnosis
58	MOD-RES	Binary	Malignant/benign	0.89	F-score = 0.89	BC detection
59	DCNN	Binary	Malignant/benign	0.99	F-score = 0.99	BC detection
60	MK-SVM	Multiclass	Normal/malignant/benign	0.96	Precision = 0.94	BC image classification
61	Kernel-SVM	Binary	Malignant/benign	0.99	–	BC detection
62	SVM, ResNet50	Binary	Malignant/benign	0.74	AUC = 0.82	BC detection
63	MobileNet	Binary	Malignant/benign	0.86	Sensitivity = 0.93	BC detection
64	SCNN, DCNN	Binary	Malignant/benign	0.951	F-score = 0.957	BC diagnosis
65	IMPA-ResNet50	Binary	Malignant/benign	0.98	AUC = 0.9788	BC diagnosis
66	ResNet50	Binary	Malignant/benign	–	AUC = 0.866 ± 0.015	Breast lesion classification
67	SVM, KNN, DT, LDA	Binary	Malignant/benign	0.963	Sensitivity = 94.1% Specificity = 98.2%	BC detection

(continued)

Table 1. Continued.

Paper reference	Models/algorithm	Binary or multiclass	Classes	Accuracy	Other performance evaluation parameters	Anomaly application/task
68	SqueezeNet, FSVM	Binary	Malignant/benign	0.98	F-score = 97.87%	BC detection and classification
69	EDNN, MSVM	Multiclass	Normal/benign/malignant	96.72%	Recall = 96.24%	BC detection
70	SVM, DT, RF, NB	Binary	Malignant/benign	0.99	–	BC detection
71	2D V-net 64 CNN	Binary	Malignant/benign	0.98	Sensitivity = 0.96 Specificity = 0.96	BC detection
72	AlexNet, VGG, GoogleNet CNNs	Binary	Malignant/benign	98.80%	Sensitivity = 99.62%	Breast lesion diagnosis
73	LASSO regression	Binary	Malignant/benign	–	AUC = 0.940	BC diagnosis
74	ANN	Binary	Malignant/benign	100%	Specificity = 100% Sensitivity = 100%	BC classification
75	Classification ML	Binary	Cancer or non-cancer	–	Sensitivity = 100%	BC diagnosis
76	LR	Binary	Negative and positive	–	AUC = 0.742 Sensitivity = 0.783 Specificity = 0.630	BC ALN detection
77	VGG16, SVM	Binary	Malignant/benign	–	AUC = 0.876 ± 0.031	BC detection
9	Mask RCNN	Binary	Malignant/benign	–	AUC = 0.89 Sensitivity = 0.93	Breast AD diagnosis
78	QNN, CCNN	Binary	Malignant/benign	–	–	BC detection
79	DT, ANN	Binary	Malignant/benign	93%	Sensitivity = 95%	Breast microcalcification diagnosis

LIBSVM, a library for support vector machines; AUC, area under the curve; BC, breast cancer; SVM, support vector machine; ELM, extreme learning machines; DL, deep learning; SAE, stacked autoencoder; dANN, deep learning with artificial neural network; DCNN, deep convolutional neural network; DNN, deep neural network; LASSO, least absolute shrinkage and selection operator; KNN, k-nearest neighbour; B:SWIMS, bootstrapped stagewise model selection; ROC, receiver operating characteristic; LDA, linear discriminant analysis; RF, random forests; LR, logistic regression; CNN, convolutional neural network; VGG16, visual geometry group 16; R-CNN, region-based convolutional neural network; NB, naïve Bayes; ANN, artificial neural network; DT, decision tree; VGG19, visual geometry group 19; QGA, quantum genetic algorithm; AlexNet, Alex network; VGGNet, visual geometry group network; RBFNN, radial basis function neural network; SRG, seeded region growing; NSEA, novel spectral extraction algorithm; MOD-RES, modified version of residual network 50; MK-SVM, multi-kernel support vector machine; ResNet 50, residual network 50; MobileNet, mobile network; SCNN, shallow convolutional neural network; IMPA, improved marine predators algorithm; SqueezeNet, squeeze network; FSVM, fuzzy support vector machine; EDNN, ensemble deep neural network; MSVM, multiclass support vector machine; CNNs, convolutional neural networks; ML, machine learning; ALN, axillary lymph node; AD, architectural distortion; QNN, quantum neural network; CCNN, classical convolutional neural network.

Ultrasonography

Ultrasonography is most frequently imaging technique used to screen for early-stage BC. US images are typically preferred as they, unlike MG, are not associated with radiation or compression. Nevertheless, its SEN is similar to digital MG (DM) and multiple studies confirmed its enhanced identification of invasive and node-negative BC.[80,81] Breast US (BUS) images are known to reliably delineate between b-BT and m-BT in five distinct areas, namely, shape, orientation, margin, echo patterns and posterior acoustic profiles.[82] Despite the aforementioned advantages, US images can have some major shortcomings, namely, reduced resolution, contrast and blurred edges owing to noise originating from speckle and acoustic shadowing. Therefore, elucidation and BC identification from BUS is often challenging and operator dependent. CAD systems are an alternative automated approach for BC classification and it minimizes dependency on the operator.[83] BUS SEN and SPC can potentially be further augmented using ML. ML serves as a second reader that assists radiologists in forming a diagnosis.[84,85] ML algorithms study profiles of known cases to generate models that accurately diagnose patients with undetermined diseases. LR and naïve Bayes are two such programs with distinct learning capacities, and they show similar BC diagnosis on sonographic images.[86] These programs were integrated to generate an augmented model, which was grayscale profile-trained only with serial adaptive boosting.[84] Rather than boosting homogenous weak learner like canonical AdaBoost, the integrated program boosts heterogeneous classifiers like naïve Bayes and LR which have established performances, and which closely agree on individual cases.[84–86]

Given that the BUS and elastography ultrasound (EUS) integration automatically provides more data for BC diagnosis, the bimodal US-based CAD has generated much attention in recent times. A previous study reported the application of a deep polynomial network on dual modal BUS and EUS properties to delineate between m-BT and b-BT tumours.[87] Another study also employed pretrained bi-channel CNNs to study and integrate bimodal US image profiles for BC tumour detection.[88] Although the aforementioned studies indicate a strong efficacy of bimodal tools, the single-mode BUS-derived CAD offers more flexibility and broader application, and its performance can be further enhanced with transfer learning using EUS as the source domain.

Artificial intelligence, composed of ML and brain-inspired DL neural networks, have great potential in combating current challenges within the global healthcare system. Recent AI applications in medical US images include numerous specific tasks, such as image segmentation and biometric measurement.[8] Prior investigations successfully established tools for automated BT segmentation as well as CAD US intranodal vascularity quantification.[89] AI complements radiomics, which is used to extract meaningful imaging information, such as textural and wavelet data, which cannot otherwise be acquired by humans. Using the aforementioned radiomics profiles, one can train AI to conduct its own diagnosis, for example, stratifying a tumour as b-BT or m-BT. The precise identification of b-BT or m-BT from US imaging is critical for the timely intervention of BC patients. Moreover, effective decision support systems can markedly enhance radiological diagnostic ability.

A retrospective investigation evaluated US imaging and clinical information from 140 surgically confirmed BC cases.[90] In particular, the study examined twelve US and colour Doppler images using ML tools, among which eight profiles were statistically different between the two images ($P < 0.05$).[90] The final diagnostic exhibited an AUC of 0.88, SEN of 86.96% and SPC of 82.91%.[90] Another study established an automated BC diagnostic system with enhanced precision that could be readily run on smartphones.[91] Upon US report entry, the program automatically analyses and detects lesions from all uploaded images.[91] The aforementioned process consists of three subsystems.[91] Firstly, noise is minimized within the images, followed by the reconstruction of high-quality images.[91] Subsequently, the initial subsystem is generated based on a stacked denoising autoencoder framework and generative adversarial network.[91] Then, the image is stratified according to malignancy or nonmalignancy.[91] DCCN is used to isolate significant profiles from the image.[91] Lastly, system performance anomalies are examined and eliminated, which further diminishes the false negative rate.[91] The current review extracted and organized the data in tabular form and summarized the application of US images in breast cancer diagnosis (Table 2).[6,8,80,83,90,92–118]

MRI

Breast MRI is frequently employed for the presurgical assessment of malignancy status, as well as the identification of potential ipsi- or contralateral BT. MRI-identified surplus BTs are BTs detected on presurgical MRI, which were undetected in prior MG or US. A secondary or targeted US is typically conducted to further assess the surplus BT. But, US is generally nonspecific for malignant tissue identification, and the MRI and US association rates are often widely variable between 23%–89%, based on multiple factors, including, radiologist performance or patient-specific differences.[119]

Researchers utilized deep transfer learning CAD-based diagnosis to identify BC.[120] In particular, they used multi-parametric MRI (mpMRI) relative to a simple independent CAD.[120] They demonstrated that the mpMRI tool provided superior results in delineating b-BT from m-BT; and the dynamic contrast-enhanced (DCE) image precision was 85%.[120] Nevertheless, there are certain limitations to this method.[120] Another study utilized 4-dimensional MRI by recording the maximum intensity projection from two distinct data, namely, image and feature, within a CNN via SVM classifier.[121] Based on their analysis, the image level exhibited a 91% precision, whereas, the feature level achieved a 93% accuracy.[121] However, their dataset was not generalized and did not include clinically sound information.[121]

A multiparametric radiomics model combining DCE- and diffusion-weighted imaging-extracted profiles revealed an optimal AUC (0.85; 95% confidence interval, 0.77, 0.92) and diagnostic precision (81.7%; confidence interval, 73.0, 88.6).[122] Therefore, radiomics analysis combined with multiparametric MRI ML provides an enhanced assessment of suspicious augmenting breast tumours that are indicated for biopsy on clinical breast MRI.[122] This facilitates highly precise BC detection while minimizing needless b-BT biopsies.[122]

Newly developed imaging techniques, such as US, MG, computed tomography (CT), PET and MRI are frequently employed for early BC detection. Among them, however, MRI ranks first in terms of patient prognosis, diagnostic precision, staging and presurgical planning. Furthermore, MRI also exhibits enhanced SEN than MG; and MRI diagnostic results are only minimally impacted by breast density. Hence, MRI is regarded as an essential tool for BC clinical diagnosis.[123] A previous investigation achieved an AUC of 0.654 via an ML model that analysed MRI images to delineate between triple-negative breast cancer and remaining subtypes.[124] The current review extracted and organized the data in tabular form and summarized the

Table 2. Models, classes and performance for breast ultrasound image data in selected papers.[6,8,80,83,90,92–118]

Paper reference	Models/algorithm	Binary or multiclass	Classes	Accuracy	Other performance evaluation parameters	Anomaly application/task
92	ML, SL	Binary	Malignant/benign	0.957	Sensitivity = 0.912 Specificity = 0.966	BC diagnosis
83	BPANN, SVM	Binary	Malignant/benign	98.966% ± 0.527	AUC = 98.976% ± 0.473	Breast tumour classification
80	ML	Binary	Malignant/benign	–	AUC = 0.86	BC diagnosis
93	DFCN, CNN, SVM	Binary	Malignant/benign	71.9%	AUC = 0.795	BC detection
94	SVM	Multiclass	Normal/benign/malignant	94.4%	AUC = 0.98	BC detection and classification
90	ML	Binary	TN subtype/ nTN subtype	–	AUC = 0.88 Sensitivity = 86.96% Specificity = 82.91%	BC subtypes
95	FK, FS, FB, KS, KB, SB	Binary	Malignant/benign	–	$\kappa \approx 0.944$	BC detection
86	ML	Binary	Malignant/benign	–	ROC = 0.958 ± 0.013	BC detection
96	CNN	Binary	Malignant/benign		AUC = 0.9468 Sensitivity = 0.88 Specificity = 0.876	BC detection
97	HSIC, SVM, DSPTC	Binary	Malignant/benign	91.44 ± 1.51%	Sensitivity = 90.29 ± 3.17% Specificity = 92.65 ± 5.06% AUC = 0.946	BC detection
98	LR, RF, SV, XGBoost	Binary	Malignant/benign	82.1%	AUC = 0.906	BC diagnosis
99	ML	–				BC diagnosis
100	RF, LR, SVM, AdaBoost, DT	Binary	Malignant/benign	0.974	AUC = 0.97 F1-score = 0.94	Breast tumour classification
8	RF, CNN	Binary	Malignant/benign	86%	Sensitivity = 84% Specificity = 88% F1-score = 0.83	Breast lesion classification
6	DCNN	Binary	Malignant/benign	86.5%	AUC = 0.875 Sensitivity = 86.6% Specificity = 87.1%	BC classification

(continued)

Table 2. Continued.

Paper reference	Models/algorithm	Binary or multiclass	Classes	Accuracy	Other performance evaluation parameters	Anomaly application/task
101	SVM	Binary	Malignant/benign	0.939 ± 0.039	Sensitivity = 0.922 ± 0.045 AUC = 0.984 ± 0.012	BC detection
102	SVM		Malignant/benign	86.0%	AUC = 0.885	Breast lesion classification
103	DCNN	Binary	Malignant/benign	93.53%	Sensitivity = 94.42 % Specificity= 90.75 %	Breast lesion classification
104	XGBoost, RF, SVM	Binary	Malignant/benign	88.4%	Sensitivity = 90.3% Specificity = 86.7% AUC = 0.890	BC diagnosis
105	DCNN	Multiclass	Subclasses	–	–	BC subtype identification
106	NB, SVM	Binary	Malignant/benign	89.17%	AUC = 0.95	Breast tumour classification
107	KNN	Binary	Malignant/benign	70.00%	Sensitivity = 70.00% Specificity = 70.83%	BC detection and classification
108	CNN	Binary	Malignant/benign	0.8186	Sensitivity = 0.8095 Specificity = 0.8265	BC diagnosis
109	LR, DT, RF, Bagging	Binary	DCIS, IDC	–	AUC = 0.66–0.78	BC diagnosis
110	NN, LR	Binary	Malignant/benign		AUC = 0.93 Sensitivity = 100.0%	BC diagnosis
111	PNN	Binary	Negative/positive	89.72%	Sensitivity = 84.00% Specificity = 94.74%	BC LN detection
112	CKHA, VGG-16, VGG-19, SqueezeNet	Multiclass	Malignant/benign/ normal	97.09%	Sensitivity = 95.54% Specificity = 97.65%	BC Diagnosis and classification
113	LR, SVM	Multiclass	BI-RADS category 4A/4B/4C	–	AUC = 0.908	BI-RADS 4 lesion screening
114	DNN, RF	Binary	Malignant/benign	78.5%	–	BC diagnosis
115	DNN, SVM	Binary	Malignant/benign	87.1% ± 3.3%	Sensitivity = 77.4% ± 11.8% Specificity = 92.4% ± 7.2%	BC diagnosis

(continued)

Table 2. Continued.

Paper reference	Models/algorithm	Binary or multiclass	Classes	Accuracy	Other performance evaluation parameters	Anomaly application/task
116	SVM, XGBoost, RF, LR, NB, KNN, MLP, CNN, LSTM, LDA	Binary	Negative/positive	0.891	AUC = 0.952 Kappa = 0.763	BC SLN detection
117	XGBoost, CNN, ResNet	Binary	Malignant/benign	–	AUC = 0.84	BC detection
118	DL, ML	Binary	Malignant/benign	–	–	BC detection

ML, machine learning; SL, supervised learning; BC, breast cancer; BPANN, back-propagation artificial neural network; SVM, support vector machine; AUC, area under the curve; DFCN, dilated fully convolutional network; CNN, convolutional neural networks; TN, triple-negative; nTN, non-triple-negative; FK, forest-k-nearest neighbour; FS, forest-stochastic gradient; FB, forest-naïve Bayes; KS, k-nearest neighbour-stochastic gradient; KB, k-nearest neighbour-naïve Bayes; SB, stochastic gradient-naïve Bayes; ROC, receiver operating characteristic; HSIC, Hilbert-Schmidt Independence Criterion; DSPTC, doubly supervised parameter transfer classifier; LR, logistic regression; RF, random forests; SV, support vector; XG Boost, extreme gradient boosting; Adaboost, adaptive boosting; DT, decision tree; DCNN, deep convolutional neural networks; NB, naïve Bayes; KNN, k-nearest neighbour; DCIS, ductal carcinoma in situ; IDC, invasive ductal carcinoma; NN, neural network; PNN, probabilistic neural network; LN, lymph node; CKHA, chaotic krill herd algorithm; VGG-16, visual geometry group-16; VGG-19, visual geometry group-19; SqueezeNet, squeeze network; BI-RADS, breast imaging-reporting and data system; DNN, deep neural network; MLP, multilayer perceptron; LSTM, long short-term memory; LDA, linear discriminant analysis; SLN, sentinel lymph node; ResNet, residual network; DL, deep learning.

application of MRI in breast cancer diagnosis (Table 3).[109,119,120,123,125–166]

Others

X-ray mammography is often employed for BC identification. However, it is a highly invasive procedure because X-rays damage tissues and quite frequently fails to determine the tumour size. Lately, thermography has emerged as a safer non-invasive approach, with no contact imaging. This process does not involve ionizing radiation, venous access or other invasive protocols. Thermography used human body-emitted infrared electromagnetic radiation that is picked up by a thermographic camera for analysis by a CAD system. However, for widespread application, it is imperative to enhance the accuracy of these new tools. ML has successfully enhanced diagnostic precision while minimizing the presence of false positives and false negatives while analysing breast thermograms.

Various forms of research have examined thermography-based BC identification. This current review will present recent significant publications. A study published in 2023 reported a new AI- and thermography-based CAD system that assists radiologists in accurately diagnosing breast diseases.[167] The procedure for this new tool is as follows: using the U-net model, an intersection over an 89.03% union is achieved.[167] The segmented thermograms then undergo textural assessment and vascular network analysis to isolate significant profiles.[167] Subsequently, via implementation of the supervised learning algorithm-based classifiers and usage of the retrieved profiles, the normal versus abnormal thermograms are identified.[167] This process of BC identification was further confirmed as highly effective, revealing optimal stratification while using SVM, with a 94.4% accuracy, 96.2% precision, 86.7% recall, 91.2% F1-score and 98.3%

Table 3. Models, classes and performance for breast magnetic resonance imaging data in selected papers.[109,119,120,123,125–166]

Paper reference	Models/algorithm	Binary or multiclass	Classes	Accuracy	Other performance evaluation parameters	Anomaly application/task
125	ML	Binary	Malignant/benign	81.5%	Specificity = 91.4%	BC detection
119	SVM, DT	Binary	Malignant/benign	86.7%	ROC = 91.1%	BC additional lesion detection
122	SVM	Binary	Malignant/benign	81.7%	AUC = 0.85	BC detection
126	SVM, KNN, RF	Binary	Malignant/benign	78.50	AUC = 78.50 Sensitivity = 0.923 Specificity = 0.722	BC diagnosis
127	SVM, LR, NB, KNN	Binary	Malignant/benign	0.93	Sensitivity = 0.85 Specificity = 0.89 AUC = 90.9%	BC diagnosis
128	LR	Binary	Malignant/benign	–	AUC = 95.25	BC diagnosis
123	SVM-RFE, RUSBoost	Binary	Malignant/benign	–	AUC = 0.9617	BC diagnosis
129	DCNN, VGGNet	Binary	Malignant/benign		AUC = 0.88	Breast lesion classification
130	RF	Binary	Malignant/benign	–	ROC = 0.852	Breast lesions classification
131	DT	Binary	3+IHC/FISH+	83.9%	Sensitivity = 86.5% Specificity = 80.0%	HER2 expression level
132	SVM, Bayes, KNN, RF, DT	Binary	PCR/nonPCR	–	AUC = 0.879 Specificity = 82.19% Sensitivity = 83.57%	BC NACT Efficacy
133	RF, LR, NB	Multiclass	Luminal A/luminal B/ HER2-enriched/ triple-negative	–	AUC = 0.75	BC phenotype
134	SVM	Binary	Malignant/benign	74.1%	AUC = 0.77 ± 0.06	BC diagnosis
120	CNN, SVM	Binary	Malignant/benign	–	AUC = 0.87	BC diagnosis
135	SVM	Binary	Malignant/benign	–	AUC = 0.87	BC diagnosis
136	SVM	Binary	Malignant/benign	–	AUC = 0.925	BC diagnosis
137	IsoSVM	Binary	Malignant/benign	–	Sensitivity = 82.5% Specificity = 80.5% AUC = 0.87	BC detection

(continued)

Table 3. Continued.

Paper reference	Models/algorithm	Binary or multiclass	Classes	Accuracy	Other performance evaluation parameters	Anomaly application/task
[138]	U-Net CNN	Binary	Malignant/benign	98.97 ± 0.13	Sensitivity = 96.20 ± 0.82 Specificity = 99.54 ± 0.10	BC detection
[139]	LR, ML	Multiclass	TP/FP/negative	–	AUC = 0.50–0.53	Preoperative Breast detection
[140]	DCNN, AdaBoost	Multiclass	Malignant/benign/normal	97.2%	Sensitivity = 98.3% Specificity = 96.5%	BC detection
[141]	KNN, DT, NB, RF, SVM	Binary	Malignant/benign	83.67%	–	BC diagnosis
[142]	RF, LR, GNB, LDA, SVM, MLP	Multiclass	HR+/HER2−/HER2+/TNBC	0.735	AUC = 0.896	BC subtype identification
[143]	DT	Binary	PD-L1+/ PD-L1−	88.2%	Sensitivity = 90.7% Specificity = 85.1%	BC PD-L1 expression status
[144]	LR, SVM, RF	Binary	Complete responders/ non-complete responders	–	AUC = 0.91–0.98	BC neoadjuvant chemotherapy response
[145]	KNN, RF	Binary	Malignant/benign	94.1%	Sensitivity = 99.0% Specificity = 87.8%	Breast tumour classification
[146]	RF, SVM, PCA	Binary	Malignant/benign	–	AUC = 0.85	BC diagnosis
[147]	SVM	Binary	Malignant/benign	83.2%	–	BC diagnosis
[148]	ML	Binary	Malignant/benign	–	–	BC diagnosis
[149]	LogitBoost, RF	Binary	luminal A/luminal B	–	Sensitivity = 80.0%	BC prognosis
[150]	SVM, NB, LDA, LR	Binary	Malignant/benign	85.9%	Sensitivity = 95.3% Specificity = 71.2%	BC differential diagnosis
[151]	LDA, SVM	Binary	Malignant/benign	0.84	AUC = 0.88	BC detection
[152]	SVM	Binary	Malignant/benign	–	AUC = 0.90	BC diagnosis
[153]	SVM	Binary	Poor response/ excellent response	–	AUC = 0.75	BC neoadjuvant chemotherapy Response
[154]	Inception-v3, VGG-16, ResNet50, Inception-ResNet-v2	Multiclass	ER–/+/PR–/+/ PAM50 subtypes	0.742	AUC = 0.942 AUC = 0.920	BC subtype identification
[155]	SVM	Binary	Malignant/benign	88.5%	AUC = 0.96	Breast lesion diagnosis

(continued)

Table 3. Continued.

Paper reference	Models/algorithm	Binary or multiclass	Classes	Accuracy	Other performance evaluation parameters	Anomaly application/task
[109]	LR, DT, RF, Bagging	Binary	DCIS/IDC	–	AUC = 0.66–0.78	BC diagnosis
[156]	CNN-SVM	Binary	Malignant/benign	95.28%	AUC = 0.974	BC classification
[157]	CNN	Binary	Normal/abnormal	88.02%	AUC = 0.880	BC LNM detection
[158]	SVM	Binary	Malignant/benign	–	AUC = 0.983	BC diagnosis
[159]	NB, RF, AB, DT, KNN, SVM, LDA, LR	Binary	Low/high	–	AUC = 0.79	BC Ki-67 and histological grade detection
[160]	SVM, RF, XGB	Binary	Phyllodes tumours/ fibroadenomas	93.0%	AUC = 0.97 sensitivity = 92.0% specificity = 94.0%	Breast lesion differential diagnosis
[161]	RF, DT, SVM	Binary	Malignant/benign	0.88	AUC = 0.95	BC diagnosis
[162]	VGG-16, KNN, XGBoost, LightGBM	Binary	Negative/positive	–	AUC = 0.90	BC LNM detection
[163]	RF	Binary	PCR/non-PCR	80.0%	AUC = 0.77	BC neoadjuvant chemotherapy Response
[164]	LR	Binary	Negative/positive	–	AUC = 0.76	BC recurrence detection
[165]	NB	Binary	Malignant/benign	67%	Sensitivity of 70% Specificity of 66%	BC diagnosis
[166]	SVM, RF, DT, NB, LR, LDA, MLP	Binary	Negative/positive	0.82	AUC = 0.85	TNBC TP53 mutation status

ML, machine learning; BC, breast cancer; SVM, support vector machine; DT, decision tree; ROC, receiver operating characteristic; AUC, area under the curve; KNN, k-nearest neighbour; RF, random forests: LR, logistic regression; NB, naïve Bayes; SVM-RFE, support vector machine-based recursive feature elimination; RUSBoost, random undersampling boosting; DCNN, deep convolutional neural networks; VGGNet, visual geometry group network; IHC, immunohistochemistry; FISH, fluorescence in situ hybridization; HER2, human epidermal growth factor receptor 2; PCR, pathological complete response; NACT, neoadjuvant chemotherapy; CNN, convolutional neural network; IsoSVM, isomap support vector machines; U-Net, U-network; TP, true positive; FP, false positive; AdaBoost, adaptive boosting; GNB, Gaussian Naïve Bayes; LDA, linear discriminant analysis; MLP, multilayer perceptron; HR, hormone receptors; TNBC, triple negative breast cancer; PD-L1, programmed death receptor ligand 1; PCA, principal component analysis; VGG-16, visual geometry group-16; ResNet50, residual network 50; ER, oestrogen receptor; PR, progesterone receptor; PAM50, prediction analysis of microarray 50; DCIS, ductal carcinoma in situ; IDC, invasive ductal carcinoma; CNN-SVM, convolutional neural networks-support vector machine; LNM, lymph node metastasis; AB, adaptive boosting; XGB, extreme gradient boosting; XGBoost, extreme gradient boosting; GBM, gradient boosting machine.

true negative rate.[167] Another study conducted a binary stratification of m-BT and b-BT using breast thermographic images.[168] The authors employed 94 images (320×240) with biopsy-confirmed diagnoses, among which, 60 exhibited m-BT and 34 exhibited b-BT.[168] The authors used three distinct image analytical tools, namely, blinded screening mode (SBS), clinical assessment and ANN.[168] The first method provided a risk score ranging from 0 (minimum risk) to 7 (very high risk).[168] The remaining methods yielded a binary result, identifying whether a given lesion was m-BT or b-BT.[168] The analyses revealed that the ANN method excelled over the others, with a 81.8% accuracy, relative to 66.7% for SBS and 71.4% for clinical analysis.[168] Another study reported using CNN that included data augmentation and a fine-tuning optimization algorithm and a an automatic BC diagnosis.[169] These received a 92% accuracy and demonstrated that data augmentation considerably enhanced tumour stratification in breast thermography, particularly when data were scarce.[169]

Furthermore, among studies examining thermographic images for BC classification, one demonstrated exhibited an SEN of 0.812 and an SPC of 0.882.[170] Additionally, screening tomosynthesis also garnered much attention owing to its enhancement of cancer detection rates, along with diminished false positive rates.[171] Similarly, a study evaluated screen-identification approaches for tomosynthetic single-reading versus double-reading mammograms, and demonstrated an 8.2 versus 6.3 cancer detection rate per 1000 screens.[172]

Additionally, there is also BT identification using highly advanced microwave systems, which facilitate a much safer, non-ionizing approach to delineate between healthy and non-healthy tissues, based on individual dielectric profiles. The present microwave breast imaging research can be categorized as follows: microwave tomography (MT) and ultra-wideband (UWB) radar techniques. MT utilizes antennas with a matching liquid, whereas UWB employs 60 antennas with a matching liquid.[173]

More recently, ML approaches have been extensively examined for detecting BTs. In addition, DL methods have also been extensively examined. Currently, for BT microwave imaging, ML and DL have been used to analyse microwave datasets from numerical simulations or phantoms measurements.[174] Research evaluated for the first-time cancer detection using UWB enhanced by ML and conventional breast examinations.[174] The authors demonstrated that the SVM quadratic kernel classified breast information with 98% precision.[174] The current review extracted and organized the data in tabular form and summarized the application of CT, breast thermal imaging, PET and microwave imaging technology in the diagnosis of breast cancer (Table 4).[92,109,147,149,151,167,174–187]

Conclusion

In conclusion, the use of ML techniques for BC imaging has the potential to improve the accuracy and efficiency of diagnosis, classification and prediction, thereby improving patient outcomes and reducing healthcare costs. ML still has its limitations, both in terms of imaging and pathological diagnosis, and ML cannot make a diagnosis of untrained diseases. In addition, ML requires a large amount of training data support, but because of research confidentiality or patient privacy protection, the effectiveness, safety and universality of data are the key issues troubling the clinical application of ML. However, further research is needed to address the challenges and limitations of these techniques, and to develop standardized protocols and benchmarks for evaluating their performance.

Table 4. Models, classes and performance for breast thermographic techniques, positron emission tomography and other combined examination data in selected papers.[92,109,147,149,151,167,174–187]

Paper reference	Relevant examinations	Models/algorithm	Binary or multiclass	Classes	Accuracy	Other performance evaluation parameters	Anomaly application/task
175	CBCT	RF, KNN, BPN, SVM	Binary	Malignant/benign	–	AUC = 0.91 Sensitivity = 0.85 Specificity = 0.82	BC detection
176	PET	SVM	Binary	Malignant/benign	0.85	AUC = 0.89 Sensitivity = 0.94 Specificity = 0.77	BC diagnosis
177	PET	CNN, MLP	Binary	Malignant/benign	–	AUC = 0.947	BC diagnosis
149	PET	LogitBoost, RF	Binary	Luminal A/Luminal B	–	AUC = 75% Sensitivity = 88.0%	BC subtypes
178	PET	MultiResUnet3D	Binary	Negative/positive	–	AUC = 0.775	BC NAC response
179	Thermography	CNN, ELM, SVM, RF, MLP	Binary	Malignant/benign	0.8843	AUC = 0.9675	BC detection
180	Microwave	RF	Binary	Malignant/benign	0.87	–	BC diagnosis
181	PS-OCT	SVM	Multiclass	Malignant/fibroadipose/ stroma	93.5%	–	Breast tissue types
174	UWB	MLP-NN, SVM, KNN	Binary	Lesion-containing/ lesion-free	98%	Sensitivity = 0.97 Specificity = 0.99	Breast lesion detection
167	Thermography	DNN, SVM	Binary	Normal/ abnormal	94.4%	F1-score = 91.2%	BC Detection
182	PAT	SVM, AlexNet, GoogleNet	Binary	Malignant/benign	0.9118	AUC = 0.8143 Sensitivity = 0.8571	BC diagnosis
183	DCE-MRI, MGs	RBFNN, KNN, SVM, RF	Binary	Malignant/benign	–	AUC = 99.10%	BC detection
147	DCE-MRI, MG	SVM	Binary	Malignant/benign	83.3%	Specificity of 82.1%	BC diagnosis
109	MG, US, MRI	LR, DT, RF, Bagging	Binary	DCIS/ IDC	–	AUC = 0.66–0.78	BC diagnosis
184	MG	RF, SVM, LMT, NB, KNN	Binary	Malignant/benign	–	Wins = 83%	BC detection
185	Pathological images	deep learning, CNN, InceptionResNetV2	Binary	Cancerous/ non-cancerous	91%	–	BC risk detection
186	US	XGBoost Tree, LR	Binary	Malignant/benign	–	AUC = 0.90 Sensitivity = 100%	Breast masses classification
92	US	SL, ML	Binary	Malignant/benign	95.7%	Sensitivity = 0.912 Specificity = 0.966	BC diagnosis

(continued)

Table 4. Continued.

Paper reference	Relevant examinations	Models/algorithm	Binary or multiclass	Classes	Accuracy	Other performance evaluation parameters	Anomaly application/task
187	X-rays, US, MI, PET, EIT	PSOWNN	Multiclass	Malignant/benign/normal	98.6%	Specificity = 98.8%	BC detection
151	CEM, DCE-MRI	LDA, SVM	Binary	Malignant/benign	0.84	AUC = 0.88	BC detection

CBCT, cone-beam computed tomography; RF, random forests; KNN, k-nearest neighbour; BPN, back propagation neural networks; SVM, support vector machine; AUC, area under the curve; BC, breast cancer; PET, positron emission tomography; CNN, convolutional neural network; MLP, multilayer perceptron; NAC, neoadjuvant chemotherapy; ELM, extreme learning machines; PS-OCT, polarization-sensitive optical coherence tomography; UWB, ultra-wideband; MLP-NN, multi-layer perceptron neural network; DNN, deep neural network; PAT, photoacoustic tomography; DCE-MRI, dynamic contrast enhancement-magnetic resonance imaging; MGs, mammographic images; RBFNN, radial basis function neural network; MG, mammography; US, ultrasonography; MRI, magnetic resonance imaging; LR, logistic regression; DT, decision tree; DCIS, ductal carcinoma in situ; IDC, invasive ductal carcinoma; LMT, logistic model trees; NB, naïve Bayes; XGBoost, extreme gradient boosting; SL, supervised learning; ML, machine learning; MI, microwave imaging; EIT, electrical impedance tomography; PSOWNN, particle swarm optimized wavelet neural network; CEM, contrast-enhanced mammography.

Author contributions

Wenjie Shi and Qing-Qing Yu conceived of the presented idea, drafted and reviewed the relevant literature. Leilei Yuan, Weidong Chen and Haibo Zhao contributed to the design of this study. Ya Guo and Heng Zhang contributed to data collection. All authors have read and agreed to the published version of the manuscript.

Declaration of conflicting interest

The authors declare that there are no conflicts of interest.

Funding

This work was supported by the Key R&D Program of Jining (no. 2023YXNS051).

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