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Review

Recent Advances In Breast Cancer And Role Of Artificial Intelligence In Prediction And Diagnosis

Dr. Mekala Anusha^{*1}, Dr. Vummarao Tejaswi², Vangari Akanksha³, Vasinni Sravani³, Yash Desai³, Zoya Fatima³, Anuraj Malhari Surve³ Muvvala Sudhakar⁴



^{1,2}Assistant professor, Malla reddy college of pharmacy, Maisammaguda, Dhullapally, Secunderabad, 500100

³Student, Malla reddy college of pharmacy, Maisammaguda, Dhullapally, Secunderabad, 500100

⁴Professor, Malla Reddy College Of Pharmacy, Maisammaguda, Dhulapally, Secunderbad, 500100

*Author for Correspondence: Dr. Mekala Anusha

Email: anushayadav010@gmail.com

	Abstract
Published on: 24 Jun 2025	<p>Breast cancer is still the most common cancer in women around the world, but research in biology, screening, and treatment is moving quickly. In the last five years, progress in molecular biology (like genomic profiling and the development of targeted therapies) and detection technologies (like 3D imaging) have made care more personalized and improved outcomes. At the same time, artificial intelligence (AI), which includes traditional machine learning, deep learning, and radiomics, has become a powerful tool for predicting and diagnosing breast cancer. AI can find patterns in large imaging and molecular datasets that are too small for people to see. AI-driven algorithms have been shown to work as well as or better than human experts in breast imaging techniques like mammography, ultrasound, MRI, digital breast tomosynthesis, and thermography, as well as in digital pathology (1,2). We look at these changes from both a global and an Indian point of view: India has a lot of problems because the disease starts at a young age. This has led to new screening methods like AI-enabled thermography. We also talk about issues with data quality, how to make it fit into clinical workflow, ethics and regulations, and we give an overview of where we want to go in the future with clinical deployment. This review is set up to talk about (1) new developments in breast cancer biology and treatment, (2) traditional and AI-enabled detection and screening, (3) how AI is used in certain imaging and pathology methods, (4) how AI compares to traditional methods, (5) problems and future directions, with a focus on global statistics and the Indian experience.</p>
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INTRODUCTION

Breast cancer rates and deaths have gone up all over the world. There were about 2.3 million new cases of female breast cancer diagnosed around the world in 2022 (3), making it the most common cancer in women. It killed about 670,000 people in 2022 (4). Big epidemiological studies show that breast cancer rates are going up, especially in low- and middle-income countries (LMICs). Breast cancer is a problem for women in India at younger ages: it makes up about 13–28% of all female cancers in India (about 216,000 cases in 2022). (5 6). An average age of onset is near 50 years. Indian women are classified as diagnosed in comparatively advanced stages when compared to their Western counterparts. Advancements in early detection, imaging, and therapy have begun to change the outlook for this disease. Concurrently, AI comprising machine learning (ML), deep learning (DL), and radiomics has found its way into breast cancer research and care. The AI systems learn from imaging (mammograms, ultrasound, MRI, digital pathology slides) to identify lesions, classify, or stratify for risk(1 2). This review surveys the most important advances that have taken place over the last five years in areas of breast cancer biology, screening, and treatment before turning to AI-driven prediction and diagnosis. We emphasize the international scene and present selected Indian developments, including a description of new AI-based screening initiatives. Finally, we contrast AI-based techniques with traditional methods and discuss practical challenges of clinical implementation.

Advances in Breast Cancer Biology and Treatment

Thanks to new research in the molecular heterogeneity of breast cancer, our understanding has significantly increased. High-throughput genomic-meets-multi-omics (genomics, transcriptomics, proteomics) focused on understanding subtypes and pathways possible for targeted therapy (7). For instance, gene-expression profiling (PAM50) further crystallizes luminal versus basal-like subdivision and directs treatment regimens accordingly. With new therapies evolving from germline and somatic mutations on a daily basis, the molecular subtype classification is losing relevance as of therapy. There is now an explosion of targeted therapies: PARP inhibitors (Olaparib, talazoparib) for BRCA-mutant breast cancers show major benefits in clinical trials (8). HER2-targeted therapies are now provided by novel antibody-drug conjugates (trastuzumab–deruxtecan) to improve outcomes even in HER2-low tumors. CDK4/6 inhibitors (palbociclib, ribociclib, abemaciclib) have become standard in the advanced hormone-receptor positive (HR+) disease setting and more recently as adjuvant treatments. Immune checkpoint inhibitors (e.g. pembrolizumab) have been approved in high-risk triple-negative breast cancer, further responding to the emergence of immunotherapy. Endocrine therapies have recently evolved as well: novel oral selective estrogen receptor degraders (SERDs) such as elacestrant are now available for patients with ESR1-mutant disease, indicating progress to overcome endocrine resistance.

In association with therapy, screening and detection means have evolved too. Digital breast tomosynthesis (DBT) is a form of three-dimensional (3D) mammography, with higher sensitivity, especially in dense breasts (8). Magnetic resonance imaging (MRI), known well for those at high-risk, offers highly sensitive detection of breast cancer. Contrast-enhanced spectral mammography (CESM), a newer form of imaging offers highlights of tumor angiogenesis. Liquid biopsies (circulating tumor DNA) currently undergoing evaluation for early detection and recurrences, are still investigational.

Medicine in India has many of the same public health challenges. India's national program endorses performing clinical breast examinations (CBE) on women in the 30–65 age group, stating, "Mammogram screening is encouraged starting at 40 to 50 or earlier in women with breast cancer in the family." Unfortunately, screening by mammography is low. State-level initiatives in India have piloted out-of-the-box thinking. A good example is Punjab's provincial government, which funded a measure using Thermalytix - a system of AI-enabled infra-red thermography, because it is relatively inexpensive - through pilot projects that allowed for the screening of 15,069 women aged 30 (8) . The system becomes an AI-based triage system, which calculates risk and identifies those needing imaging. These examples are another positive step forward towards using low-cost AI-enabled systems in resource-limited settings.

Advances in Detection and Screening

Screening Efficacy

A population's screening engagement remains mammography - as opposed to CT, MRI, etc. or other imaging tests; and studies indicate that mammography affords a 20–40% decrease in breast cancer death when followed organized program (9). Digital mammography has effectively replaced analog mammography as it provides better imaging quality, and viewed as a gold-standard modality. Digital breast tomosynthesis (DBT) or "3D mammography" also provides a higher standard for cancer detection with numerous studies noting better detection and lower recall rates within DBT compared to 2D mammography (9). In numerous high-resourced regions and communities, DBT or 2D+DBT, has85 been communicated to screening women. For Indian women who tend to have dense breasts and present with breast cancer at less than stage II, mammography alone as the screening modality, limits detection sensitivity; which indicates the need for adjunct modalities that may be

beneficial. As an example, ultrasound is used as routine complementary imaging in many clinics; primarily with younger women, as it has the ability to potentially identify a tumor when the mammogram appears normal (9).

Imaging Innovation

New developments include automated whole-breast ultrasound (ABUS) and elastography technologies. The main advantage of ABUS is that it reduces operator-dependence because it provides standardized 3D ultrasound scans. There are also advancements in Breast MRI through faster sequences and new contrast protocols. Contrast-enhanced mammogram (CESM) is a unique method for detecting breast cancer encompassing mammography and intravenous contrast use to visualize tumors with enhancement. There are also increased use of radionuclide imaging (e.g. PET/CT, dedicated breast PET) for staging and monitoring therapy. These developments contribute to our armamentarium, albeit one must be mindful about the potential variability in cost and access globally.

Risk stratification

There are newer genetic- and imaging-derived risk models. We have polygenic risk scores that combine information from many variants which provide a more refined risk estimation for an individual than purely single-gene (BRCA) models can provide. MRI-based breast density and texture analyses also help refine risk estimates. In India, although there is an emphasis on awareness and self-examination due to lack of screening capability, there are some places that have started testing AI tools in order to further improve access to screening. For example, the Thermalytix study in Punjab that reported use of an AI-thermography device that was able to screen thousands of women with a median age ~41 (10) with potential cancer detection at a lower cost and without radiation. Overall, screening is more driven by technology and personalized.

Artificial Intelligence Methods: Machine Learning, Deep Learning, and Radiomics

AI includes various computational methods. Machine learning (ML) involves algorithms that learn patterns from data, such as logistic regression, support vector machines, and random forests. Deep learning (DL) is a part of ML that uses multi-layer neural networks, like convolutional neural networks (CNNs), to automatically learn hierarchical image features. Radiomics is another AI method that extracts many quantitative features, like shape, texture, and intensity, from imaging. This process turns images into "big data" for analysis. Radiogenomics combines features from imaging with genomic profiles to predict tumor biology without invasive procedures.

In practice, supervised deep learning has led recent advancements. Models are trained on labelled datasets, which consist of images with known diagnoses, to perform tasks like classification (deciding between benign and malignant), detection (finding tumors), and segmentation (defining tumor boundaries). CNNs, such as ResNet, VGG, and U-Net, are commonly used. A typical workflow includes collecting high-quality images, marking important areas, training the neural network, and validating it on separate data. Radiomics workflows also involve image acquisition, tumor segmentation, extracting features—often hundreds—and modelling with ML. By combining genomic and clinical data, these methods aim for precise diagnostics. Early studies show potential; for example, deep networks used on mammograms have reached approximately 99% accuracy in detecting masses, and radiomic models have linked MRI-derived features to estrogen receptor status. In the following sections, we will discuss AI applications in the main breast imaging techniques.

AI in Breast Imaging Modalities

Mammography

Mammography is essential for breast imaging, and AI has been widely used in interpreting mammograms. Early work focused on handcrafted features, such as texture filters, combined with machine learning. In contrast, modern approaches use deep convolutional neural networks (CNNs). CNNs can learn to tell apart benign and malignant lesions, segment masses, and classify breast density. For instance, Pillai et al. applied a VGG16 CNN to mammograms and achieved high accuracy, outperforming earlier networks. Other studies have employed CNNs to predict tumor molecular subtype, such as differentiating between Luminal A, Luminal B, HER2+, and basal-like, directly from imaging patterns.

It is important to note that AI is being compared to human radiologists. Several retrospective studies found that AI performance on screening mammograms matched or exceeded that of expert readers. In one Taiwanese study, referred to as AI-STREAM, using AI alongside traditional methods improved cancer detection rates by about 4% compared to standard double-reading. Similarly, the MASAI trial in the Netherlands reported a 20% increase in detection with AI-supported triage without an increase in false positives. These results indicate that AI can help reduce missed cancers. AI systems also help lighten the workload for radiologists. For example, an AI algorithm can flag clearly normal exams, allowing for single readings and reducing the workload from double readings without missing cancers.

However, challenges persist. CNNs may have difficulty with mammograms of very dense breasts, where tumor contrast is low. They might also respond inaccurately to imaging artifacts or features that are not cancer-

related unless they are trained carefully. The quality of training data is crucial. The INbreast dataset, a public collection of mammograms, has been commonly used for benchmarking. Using INbreast, Wang et al. reported that CNN-based systems achieved about 98.96% detection accuracy for mass lesions and nearly perfect sensitivity and specificity, at 100% and 94%.

Ultrasound

Ultrasound helps evaluate lumps and acts as an extra screening tool, especially for women with dense breasts. It relies a lot on the skills of the operator, but AI can help make interpretations more consistent. Convolutional neural networks (CNNs) trained on ultrasound images can identify lesions as either benign or malignant. For example, Han et al. found that a deep CNN achieved 90% accuracy (sensitivity 86%, specificity 96%) in distinguishing breast ultrasound masses (2). Improvements in ultrasound technology, like shear-wave elastography, which measures tissue stiffness, have also been combined with AI. Zhang et al. applied a two-layer deep network to elastography images to tell apart lesions, achieving 93.4% accuracy (sensitivity 88.6%, specificity 97.1%) (14). By merging standard ultrasound and elastography features, some deep learning models can even predict the status of axillary lymph nodes before surgery.

However, AI in ultrasound faces specific challenges. Unlike CT or MRI scans, ultrasound images can vary widely depending on the probe angle, pressure, and machine settings. As the authors point out, different operators may collect images in subjective ways, which can affect AI performance (14). Therefore, creating strong AI models requires large and varied datasets, and possibly real-time support for the sonographer. Despite these challenges, AI in ultrasound shows promise. Segmentation networks can automatically outline masses, and classification networks can act as a “second opinion” to highlight suspicious findings. In one study, an automated CNN was able to classify common lesion shapes from ultrasound, supporting the BI-RADS system.

Magnetic Resonance Imaging (MRI)

Breast MRI is very sensitive and is used for screening high-risk patients and solving problems. AI has been applied to breast MRI to make analysis easier. Techniques include automated lesion segmentation and computer-aided detection of enhancing lesions. For instance, Winkler et al. integrated a deep learning model into the PACS (Picture Archiving) system to highlight MRI slices with tumors, helping radiologists not to miss any lesion slices. Multiparametric MRI, which combines T1, T2, diffusion-weighted, and dynamic contrast sequences, provides valuable data. AI can combine these features. Radiomic analysis of MRI kinetics and texture can predict subtypes and therapy responses. Some studies have used CNNs or fully convolutional networks for segmenting tumors or analyzing background parenchymal enhancement in MRI.

Quantitative MRI features, like the apparent diffusion coefficient from DWI, can also support ML models. In neoadjuvant therapy, MRI-based ML models have been created to predict pathological complete response. For example, deep learning radiomic pipelines that analyze pre- and mid-treatment MRI have shown promise in predicting chemotherapy response. Overall, MRI-AI research indicates improvements in workflow by focusing on key slices and better tumor characterization. However, MRI is costly and not widely available, so AI tools for MRI mostly remain in the research phase. Considering MRI's strength in assessing treatment response and detecting subtle lesions, AI tools could improve accuracy, particularly in complex cases like multifocal disease.

Digital Breast Tomosynthesis (DBT)

Digital breast tomosynthesis creates a 3D image of the breast by taking X-rays from different angles. This helps in finding lesions that might be hidden by overlapping tissue. AI applied to DBT aims to automate the detection of lesions on each slice. Deep learning algorithms have been trained to analyze DBT volumes, generating a “maximum suspicion projection” that marks potential lesions (14). In a study comparing results, DL-based DBT readers performed better than radiologists in spotting subtle tumors (14). Radiomics in DBT is also developing; for example, texture features from DBT have been linked with tumor size and hormone receptor status (15).

One benefit of DBT is lower recall rates, while a downside is reduced resolution for small calcifications. AI may help address this issue by learning from large datasets. Although there are few prospective clinical trials of AI in DBT, retrospective data show promise. For example, using 2D mammography with DBT input in an AI model increased sensitivity and specificity compared to 2D alone. As DBT becomes commonplace in many screening programs, AI algorithms designed for its 3D data are likely to become more common.

Histopathology (Digital Pathology)

AI has made significant strides in the field of pathology, going beyond just imaging. With high-resolution whole-slide images of H&E-stained tissue, deep neural networks can now analyze these images for various tasks, including tumor detection, grading, and biomarker quantification. For instance, AI can automatically identify cancerous areas and metastases in lymph nodes, which helps pathologists manage their workload more efficiently. In the realm of breast cancer pathology, researchers have developed convolutional neural networks (CNNs) that

can differentiate between invasive carcinoma and benign tissue, as well as grade tumors. An AI model, for example, can accurately count mitotic figures, leading to more consistent grading. Other algorithms are designed to quantify tumor-infiltrating lymphocytes (TILs) or predict genomic markers, like the PAM50 subtype, based solely on morphology.

What's particularly exciting is that AI can sometimes predict molecular features without needing additional tests. Some models have been trained to determine ER, PR, HER2, and Ki-67 status directly from H&E images. These predictive models have shown impressive accuracy, which could lessen the reliance on separate immunohistochemistry tests in certain situations. AI is also being used to forecast patient outcomes and responses to therapy based on histologic patterns. For example, neural networks that analyze histopathology can predict how patients will respond to neoadjuvant chemotherapy.

As clinical workflows evolve, several FDA-approved digital pathology AI tools are now available. In diagnostic settings, an AI “second read” can catch missed micro metastases in sentinel nodes. A recent review highlighted AI's potential to minimize errors and reduce the workload in pathology, although it also pointed out that inconsistent slide staining and scanning quality can present challenges. Therefore, while the prospects for AI in pathology are promising, these systems require thorough validation across various laboratories. In India, the adoption of digital pathology is gradually increasing, and AI could play a crucial role in addressing the significant shortage of pathologists by helping to triage cases, such as distinguishing between normal and suspicious histology for further review.

Comparative Analysis: AI vs Traditional Diagnostics

AI-based methods are increasingly being compared to traditional interpretations. In the realm of imaging, numerous studies have shown that AI systems can match or even exceed human accuracy. Take, for instance, the AI mammography trial (AISTREAM), which demonstrated that when AI was paired with a single radiologist, the results were on par with or even better than the conventional approach of two radiologists (11). In this trial, the introduction of AI led to a 4% increase in cancer detection while slightly reducing recall rates (11). Similarly, retrospective studies have revealed that standalone AI was able to identify a significant number of cancers that radiologists had missed (16). In one screening group, an AI-CAD tool successfully flagged 5 out of 9 cancers that the original radiologist overlooked (16).

In the field of pathology, AI has also proven to be on par with experts for certain tasks. Deep learning models have achieved over 90% accuracy in classifying histologic images and spotting metastases (16). One analysis found that convolutional neural networks (CNNs) could detect invasive cancer in whole-slide images with sensitivity close to that of humans. Notably, integrated workflows that combine human expertise with AI often yield the best results: for example, radiologists who are assisted by AI typically demonstrate higher sensitivity (and sometimes even higher specificity) than when working alone, as AI can draw attention to subtle findings that might be missed by the human eye.

However, it's important to note that AI isn't always the superior option. Its effectiveness can vary based on the data it's trained on: models might struggle with populations or image types that differ from their training sets. For example, a mammography AI developed using data from a Western population might not perform as well on Indian data if there are differences in breast density. There are also trade-offs to consider: some AI systems may enhance sensitivity but at the expense of increased false positives, which can lead to unnecessary additional tests. Consequently, comparative studies often stress the importance of fine-tuning thresholds to strike a balance between sensitivity and specificity. In a systematic review, radiologists supported by AI achieved similar or even higher accuracy compared to those without AI assistance, although results varied depending on the study setting (18).

Challenges, Limitations, and Future Directions

Data quality and bias: AI models thrive on large, well-annotated datasets. However, differences in imaging protocols, scanner types, and patient demographics can really mess with how well these models generalize (13, 15). A lot of studies out there rely on data collected retrospectively, which means the real-world variability could be even more pronounced. Plus, it's crucial for these models to be validated across different centers. In India, for instance, gaps in infrastructure like a shortage of high-quality digital scanners and low levels of pathology digitization make it tough to gather the data needed to train robust AI systems.

Regulatory and ethical issues: the rules around AI tools in medicine are still a work in progress. In the US and EU, while some AI diagnostics have received FDA or CE approval, many are still stuck in the research phase. There are important considerations around patient consent, data privacy, and intellectual property that need clear guidelines. Ethical dilemmas also arise, such as algorithmic bias especially if the training data isn't diverse enough and transparency issues: those “black-box” deep networks can be hard to interpret, which might shake clinicians' trust. Efforts to make AI more explainable are underway, but we're not quite there yet.

Clinical implementation: AI tools need to seamlessly fit into existing workflows. This means not just integrating software (like PACS and EMR) but also ensuring that clinicians are trained to trust and effectively use AI outputs. Research suggests that AI tends to perform best as a second-reader or triage tool, rather than taking the reins on autonomous diagnoses. Keeping an eye on AI performance after deployment is another hurdle; models can “drift” if imaging practices change. Economically, there’s the balancing act between the costs of deploying AI (software, hardware, training) and the anticipated benefits (like reduced reading time and better detection). In low-resource settings, affordable AI solutions—like smartphone-based ultrasound interpretation might take precedence.

Future Directions: Looking ahead, research is really honing in on multi-modal AI, which blends imaging with clinical and genomic data to enhance prediction capabilities. Federated learning, a method that allows models to be trained on data from various institutions without the need to share raw data, could pave the way for larger datasets while keeping privacy intact. In India, there's a pressing need to keep evaluating AI screening solutions, like Thermalytix and smartphone mammography apps, to meet the demand for scalable early detection. Ultimately, we need well-structured prospective trials to truly showcase how AI can impact patient outcomes, such as reducing mortality rates. Collaborative efforts, open data sharing, and multidisciplinary teams will be essential for pushing the boundaries of AI in breast cancer care.

Global and Indian Perspectives

Breast cancer research is becoming more collaborative and data-driven on a global scale. International partnerships have rolled out extensive annotated imaging datasets, like DDSM and INbreast for mammography, and TCGA for genomics, which are essential for advancing AI technology. In Western countries, several AI-powered computer-aided detection (CAD) systems have received approval and are slowly making their way into clinics. However, the level of adoption varies; surveys show that while many radiology departments are testing AI, only a few have integrated it into their daily routines. Major medical centers are now utilizing AI tools for tasks like double-reading mammograms and digital pathology triage.

In India, the narrative around AI is closely linked to existing infrastructure challenges. Government health initiatives are looking into AI to enhance screening efforts. A notable example is the Punjab Thermalytix project, which demonstrated that an AI thermography device could efficiently screen thousands of women. Other research teams in India are working on AI algorithms for ultrasound and mammography, leveraging local datasets. For example, pilot programs have tested handheld ultrasound devices with AI interpretation in rural clinics. Oncology centers are beginning to adopt AI for tasks such as tumor segmentation on MRI scans for radiation planning. However, widespread adoption still faces hurdles: many hospitals continue to rely on analog films or offline records, and digital pathology is still in its infancy. There’s a pressing need for training programs to help radiologists and oncologists become familiar with AI tools. Crucially, any AI solution in India must prove to be cost-effective and adaptable to local conditions, including demographic variations and the lower prevalence of dense imaging.

The statistics are quite alarming: in 2020, there were around 200,000 new cases of breast cancer among Indian women, and the numbers are expected to keep climbing. With breast cancer mortality rates being higher in developing countries, enhancing early detection through AI could significantly benefit public health. Projects like the National Cancer Grid and registries led by the ICMR are gathering crucial cancer data that can help shape AI models. In short, while India does face challenges with resources, there's a vibrant community actively seeking ways for AI to overcome some of the traditional hurdles in screening and diagnosis.

DISCUSSIONS

The intersection of breast cancer research and AI technology is set to revolutionize how we care for patients. With biologically informed treatments like PARP inhibitors and immune therapies extending survival rates, AI plays a crucial role in ensuring these therapies are administered promptly by enhancing the speed and accuracy of diagnoses. For instance, AI can spot aggressive tumors during screenings that require immediate genetic testing. Innovations in radiomics and radiogenomics may soon enable non-invasive tumor phenotyping, effectively linking imaging with molecular diagnostics. The benefits of AI are already clear: higher detection rates, lighter workloads, and more reliable interpretations.

However, our review emphasizes that AI isn't a cure-all. Traditional methods like mammography and biopsy still form the foundation of patient care. AI should complement, not replace, clinical judgment. Our findings indicate that AI has improved radiologists' performance in double-reading scenarios, but we still need solid evidence connecting AI usage to long-term outcomes, such as reduced mortality rates. In lower middle-income countries, ethical implementation is vital: AI tools must be validated for the specific populations they serve and should be integrated with efforts to strengthen health systems. It's also essential to establish data privacy laws and secure patient consent for AI applications. Furthermore, interdisciplinary training is necessary so that

clinicians, data scientists, and engineers can collaborate effectively; a physician needs to grasp an AI's limitations to make the best use of its insights.

Future research is set to explore some exciting areas, like explainable AI, which helps us understand the reasoning behind a model's decisions. There's also a push to weave AI predictions into risk models that take into account lifestyle and genetic factors. We're seeing a rise in mobile health (mHealth) apps, where smartphone cameras and AI work together to analyze images or symptoms. Another promising area is longitudinal monitoring with AI, such as tracking changes in serial mammograms. Lastly, the COVID-19 pandemic has really sped up the adoption of telehealth; AI-powered teleradiology could make remote screening interpretation a reality, which would be a game-changer for countries like India that face challenges in rural healthcare access.

CONCLUSION

In the past five years, we've made remarkable strides in breast cancer research, particularly in understanding tumor biology, creating new treatments, and improving screening methods. The simultaneous growth of AI in medical imaging and pathology is opening exciting new possibilities for early detection and precise diagnosis. Machine learning and deep learning algorithms have shown impressive accuracy in analyzing mammograms, ultrasounds, MRIs, and histopathology, often matching the expertise of human specialists. AI-driven radiomics connects imaging characteristics to molecular biology, paving the way for non-invasive “virtual biopsies.” On a global scale, these advancements hold the promise of enhancing screening programs and tailoring care to individual needs. In India, for instance, innovative state-led AI screening pilots and research initiatives are customizing these technologies to fit local contexts.

That said, we need to approach the integration of AI into clinical practice with caution and a strong evidence-based foundation. It's crucial to ensure rigorous validation, maintain transparency in algorithms, and provide proper training for clinicians. We still face challenges like data diversity, regulatory approvals, and integrating these tools into existing workflows. Looking ahead, AI is poised to become a regular part of breast cancer processes whether it's flagging unusual images or predicting how patients will respond to treatment. However, its success will depend on effective collaboration between tech experts and healthcare providers. If we implement AI thoughtfully, it could significantly enhance breast cancer outcomes around the globe by enabling earlier detection, minimizing diagnostic errors, and ultimately saving lives.

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