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REVIEW

A data-driven approach to PCOS Diagnosis: Systematic review of machine learning applications in reproductive health

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Background and aim: Polycystic Ovary Syndrome (PCOS) is a prevalent endocrine disorder in reproductive-aged women, characterized by hormonal imbalances, anovulation, and metabolic abnormalities. This systematic review aims to evaluate the effectiveness, types, and diagnostic performance of ML algorithms applied in PCOS detection and classification, and to identify the most frequently used input features and methodological challenges in existing studies.

Methods: A systematic search was conducted across scholarly databased, but not limited to PubMed, Scopus, and Google Scholar for studies published between 2014 and 2024 using keywords related to PCOS and machine learning. Inclusion criteria focused on original, peer-reviewed studies applying ML models for PCOS diagnosis. Data were extracted on model type, input features, diagnostic accuracy, and study design. Quality assessment was performed using the PROBAST tool.

Results: Out of 450 identified studies, 34 met the inclusion criteria and passed the quality assessment. Supervised learning models such as Random Forest, SVM, and XGBoost showed high accuracy (up to 99%). Deep learning approaches, particularly Convolutional Neural Networks (CNNs), achieved accuracies between 95% and 99.89% in analyzing ultrasound images. Hybrid models integrating clinical and imaging data further enhanced performance. Common input features included BMI, LH/FSH ratio, AMH, and ultrasound-based ovarian morphology. However, few studies validated models on external datasets, and input feature selection lacked standardization.

Conclusion: Machine learning models such as supervised, deep learning, and hybrid approaches show strong potential in improving PCOS diagnosis by identifying complex patterns across multi-dimensional datasets. Challenges such as limited generalizability and data standardization remain, therefore future studies should focus on developing explainable ML tools, validating models in clinical settings, and leveraging diverse data types for robust, personalized PCOS diagnosis.

Keywords: polycystic ovary syndrome, machine learning, artificial intelligence, PCOS diagnosis

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Introduction

Polycystic Ovary Syndrome (PCOS) is a complex endocrine and metabolic disorder that affects reproductive-aged women. It is characterized by hormonal imbalances, irregular ovulation, and metabolic disturbances [1]. Although the exact cause of PCOS remains uncertain, it is believed to arise from a combination of genetic, environmental, and lifestyle factors [2]. Early diagnosis and personalized management are crucial to preventing long-term complications such as infertility, type 2 diabetes, and cardiovascular diseases. While medical treatments and lifestyle interventions can significantly improve quality of life, the heterogeneous nature of PCOS complicates its diagnosis and treatment [1,2,4,5].

Given this complexity, multiple diagnostic criteria have been developed, each with distinct strengths and limitations. The Rotterdam Criteria (2003), endorsed by the European Society of Human Reproduction and Embryology (ESHRE) and the American Society for Reproductive Medicine (ASRM), remain the most widely accepted. These criteria define PCOS as the presence of at least two of the following three features: oligo-or anovulation, hyperandrogenism (clinical or biochemical), and polycystic

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ovarian morphology (PCOM) on ultrasound, after exclusion of other endocrine disorders [6]. Although broad and inclusive, these criteria also encompass a wider spectrum of phenotypes, which may contribute to potential overdiagnosis in some cases [7,8].

In the era of artificial intelligence, Machine Learning (ML) models present a promising approach for enhancing diagnostic precision, by analysing large-scale datasets including hormonal, metabolic, ultrasound, and genetic information ML can identify hidden patterns not easily recognized through conventional methods. This ability supports more accurate diagnosis, risk stratification, and the design of personalized treatment plans [4,9]. Various ML algorithms, such as Decision Trees, Support Vector Machines (SVM), and Deep Learning approaches including Convolutional Neural Networks (CNNs), have demonstrated superior performance compared with traditional diagnostic methods in distinguishing PCOS from related conditions, predicting disease progression, and classifying phenotypes [7,10].

This systematic review provides a qualitative synthesis of studies exploring ML applications in PCOS diagnosis and risk assessment. Considering the heterogeneity of PCOS and the limitations of current diagnostic criteria, ML models offer the capacity to integrate diverse data types hormonal, metabolic, imaging, and genetic to uncover patterns that traditional methods may overlook. Through this review, we are exploring the effectiveness of various ML techniques reported in improving diagnostic accuracy, enhancing phenotype classification, predicting disease trajectories, and informing optimized diagnostic approaches.

Materials and methods

This systematic review was conducted according to the published protocol in Open Science Frame work- OSF (accessible through the link;https://doi.org/10.17605/OSF.IO/PDNFY) We adhered to the Preferred Reporting Items for Systematic Reviews (PRISMA) Updated Guidelines 2020 on reporting the findings. The article selection process is comprehensively illustrated in the PRISMA flowchart (Figure 1).

Search Strategy

The literature search was conducted in scholarly data bases but not limited to PubMed (National Library of Medicine), Scopus (Elsevier), and Google Scholar (Google), focusing on studies published between 2014 and 2024. Only peer-reviewed primary research articles on PCOS diagnosis using Machine Learning (ML) were considered for review. A detailed inclusion and exclusion process was applied to ensure relevance, prioritizing studies that explored ML models, AI-based diagnostic approaches, and computational methods for PCOS classification and prediction.

Boolean operators such as 'AND' and 'OR' were utilized to refine the search results, incorporating Medical Subject Headings (MeSH) terms and keywords: ("Polycystic Ovary Syndrome"[MeSH Terms] OR "PCOS") AND ("Machine Learning" OR "Artificial Intelligence" OR "Neural Networks" OR "Deep Learning") AND ("Prediction" OR "Diagnosis" OR "Modeling"). The search was restricted to open-access, full-text studies published in English within the last 10 years, ensuring access to the most recent advancements in ML-driven PCOS diagnostics.

Eligibility Criteria

The eligibility criteria for studies in this systematic review were precisely defined to ensure the inclusion of scientifically sound and relevant studies on the application of ML in PCOS diagnosis. The review considered those studies that applied ML algorithms for predicting, diagnosing, or classifying PCOS. Only fully open-access, peer-reviewed studies published between January 2014 and December 2024 were included to ensure relevance to recent advancements in AI-driven healthcare solutions.

Studies that utilized detailed and scientifically validated methodologies for applying ML techniques to PCOS diagnosis, risk prediction, or phenotyping were included. Conversely, studies published in languages other than English, studies that did not employ ML or AI models as a core analytical method, and those focusing solely on traditional diagnostic criteria without computational approaches were excluded. Conference presentations, editorials, and non-peer-reviewed articles were also excluded to maintain methodological rigour.

Data Management

The first, second, and third authors independently conducted data extraction and preliminary screening of study titles and abstracts. The fourth and fifth authors then performed a full-text review to assess eligibility based on the predefined inclusion criteria. Any discrepancies were resolved through discussion or consultation with the other authors to ensure consensus.

For each eligible study, key data were systematically tabulated, including details such as first author, year of publication, study title, study design, sample size, type of Machine Learning model used, features analyzed (e.g., hormonal, metabolic, imaging data), performance metrics (e.g., accuracy, sensitivity, specificity), and conclusions regarding the effectiveness of ML in PCOS diagnosis.

Quality assessment

The quality of the studies included in this systematic review was assessed using PROBAST (Prediction model Risk of Bias Assessment Tool). PROBAST is a widely recognized tool designed to evaluate the risk of bias (ROB) and applicability concerns in studies developing, validating, or updating predictive models in healthcare. Two authors independently conducted the quality assessment and ensured that the included studies provided robust, unbiased, and clinically relevant findings.

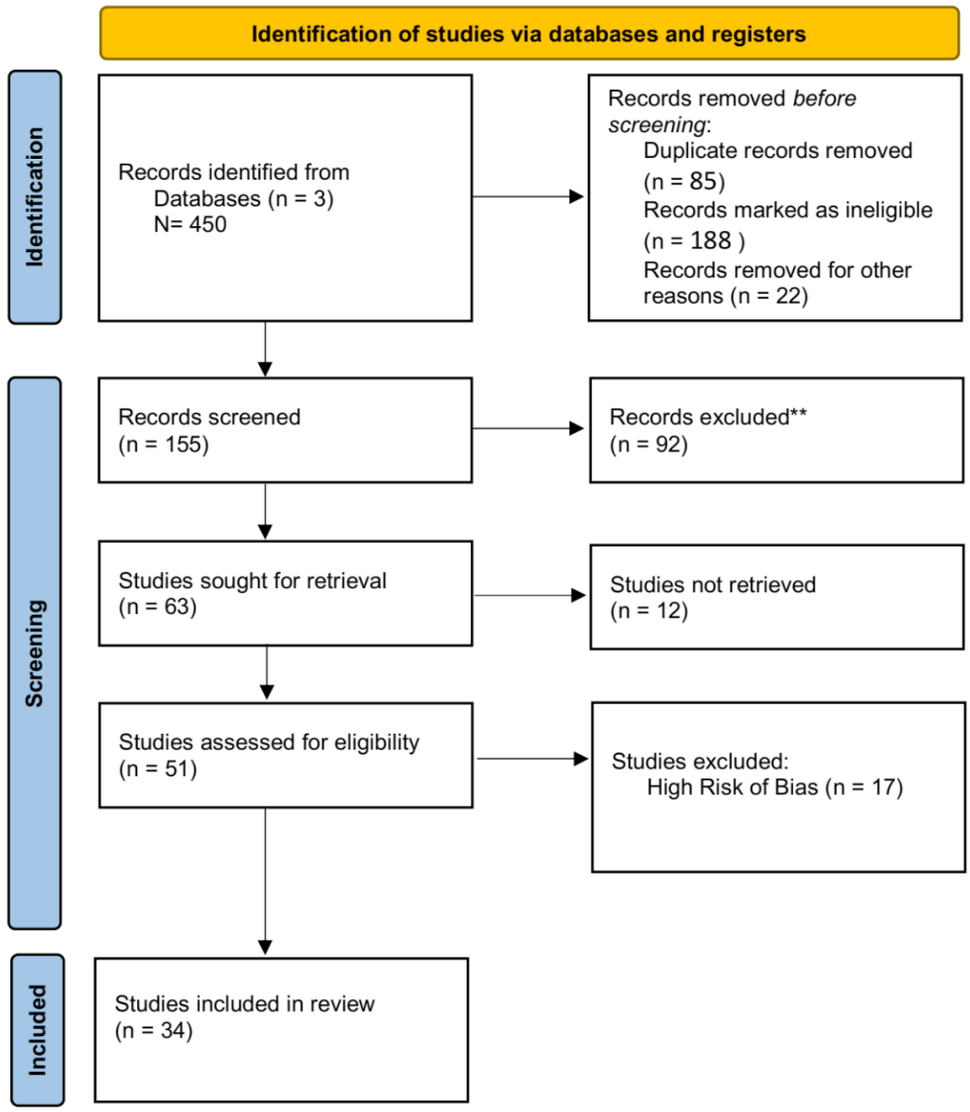

Fig. 1. Study flowchart

Results and discussion

The initial literature search across three databases yielded 450 records. After removing 85 duplicates, 188 ineligible records, and 22 for other reasons, 155 studies were screened. Following this, 92 studies were excluded based on relevance, leaving 63 for full-text retrieval. Out of these, 12 studies were not retrievable, and 51 were assessed for eligibility. Seventeen studies were excluded due to the high risk of bias, resulting in 34 studies meeting the inclusion criteria. These studies employed various machine learning techniques to investigate ML models in PCOS diagnosis and risk stratification, representing contributions from countries like Bangladesh, China, Finland and Sweden, India, Saudi Arabia, South Korea, Egypt, United Kingdom (UK), United States of America (USA) and Mexico (See Table 1).

Different ML models in PCOS prediction

The accuracy of ML models in PCOS prediction varied from 82.1% to 99.89%. Different types of ML models were used in the studies, such as Supervised Learning, Unsupervised Learning, Deep Learning, Ensemble Learning, and Hybrid Models.

Supervised Learning models

This is a type of ML where models are trained on labelled datasets, meaning the algorithm learns from input-output pairs. Logistic Regression, Random Forest, Support vector machines (SVM) and KNN are some examples of supervised machine learning models used in the studies. These models are used for predicting PCOS based on clinical and biochemical data. Among them, Random Forest and SVM perform well due to their ability to handle high-dimensional datasets and complex patterns [4,9] (Figure 2).

The logistic regression model was used as a baseline model in PCOS prediction studies and reported a moderate accuracy of 82%- 92%. Moreover, it is observed that this model works well with clinical and biochemical markers, but struggles with non-linearity in complex datasets [11,12].

In SVM, they use hyperplanes to separate data points into distinct classes with maximum margins and achieve an

Table 1. Full data extraction table of all the studies included in the review

Author, Year	Country	Type of ML	Inputs Used	Other Outcomes	Accuracy
Suha & Islam et al., 2022 [1]	Bangladesh	CNN with transfer learning and stacking ensemble ML (e.g., XGBoost).	594 ovary USG images	Reduced execution time; highest accuracy of 99.89%.	99.89%
Lv et al., 2022 [2]	China	ResNet18, U-Net with attention module, Multi-Instance Learning	721 full-eye scleral images	Highlighted the impact of PCOS on scleral blood vessel patterns.	92.90%
Sowmiya et al., 2024 [3]	India/Saudi Arabia	YOLOv8, Custom CNN (FNet), Random Forest, SGD, k-Star	Ultrasound images (100 subjects: 50 normal, 50 PCOS), additional GAN-augmented datasets.	F-Net outperformed pre-trained models and other classifiers, achieving 97.5% accuracy. YOLOv8 achieved high follicle detection precision. GAN data augmentation improved DL model performance.	95%–97.5%
Rachana et al., 2021 [4]	India	KNN	Ultrasound images of ovaries (50 affected and non-affected cases)	KNN achieved the highest classification accuracy of 97% compared to other classifiers like SVM and Decision Tree.	97 %
Luo et al., 2019 [5]	China	Logistic regression	186 subjects, including 95 PCOS patients and 91 healthy controls, with metabolic, hormonal, and cytokine data	Combining three biomarkers showed AUC of 0.90–0.93 with sensitivity of 82.1% and specificity of 92.3%.	82.1%–92.3%
Sumathi et al., 2021 [6]	India	CNN	Ultrasound images collected from online datasets (e.g., ultrasoundimages.com).	Achieved 85% accuracy using CNN. Included metrics like area, solidity, and extent for feature extraction.	85 %
Xu et al., 2022 [7]	China	Logistic Regression (LASSO)	11,720 ovarian stimulation cycles, biomarkers (AMH, BMI, UML, AND)	Achieved AUCs of 0.855–0.865 across training, validation, and testing datasets.	~
Soni et al., 2025 [8]	India/South Korea	ResNet-50, Random Forest, SVM, Logistic Regression	Ultrasound images (3200 training, 1468 testing) and clinical data (39 features)	SVM achieved the highest performance with accuracy of 99%, followed by Logistic Regression (97%) and Random Forest (95%). SHAP plots revealed critical feature contributions.	SVM: 99%, Logistic Regression: 97%, Random Forest: 95%
Vora et al., 2023 [9]	India	Optimized KNN, SVM, Neural Networks	541 patient data points, 45 features reduced to 6 key features through statistical methods	KNN achieved 99.5% accuracy; SVM also performed well. Optimized classifiers outperformed conventional approaches.	99.5 %
Bhat et al., 2024 [10]	India	Random Forest (Patient Model), CNN (Doctor Model)	Clinical data (e.g., FSH/LH values) and ultrasound scans	Random Forest achieved 90% accuracy on clinical data; CNN achieved 97% accuracy on ultrasound images.	RF:90%, CNN: 97%
Kermanshahchi et al., 2024 [11]	USA	CNN	1,932 pelvic ultrasound images (1,145 normal, 787 PCOS positive)	Achieved 100% accuracy, sensitivity, and specificity; F1 score: 0.905. High recall (100%) indicates no false negatives.	99.9 %
Zad et al., 2024 [12]	USA	Logistic regression and SVM and GBM & RF	Hormone values (FSH, LH, estradiol, and sex hormone binding globulin)	Overall predictive accuracy was high for all models	85%, 81%, 80%, and 82%, respectively in Models I, II, III and IV.
Barrera et al.,2023 [13]	USA and Mexico	Specific algorithms mentioned may include decision trees, support vector SVM, random forests, and neural networks.	Clinical data (e.g., menstrual history, BMI), biochemical markers (e.g., hormone levels), and imaging data (e.g., ultrasound findings).	High accuracy and minimal or zero false positive	89% to 100%
Lim et al., 2023 [14]	China	KNN, SVM, DT, RF, Logistic Regression, Voting, and LSTM	Radial pulse wave signal	Acceptable overall accuracy	72.174%
Elmannai et al., 2023 [15]	Saudi Arabia and Egypt	Logistic regression, RF, DT, NB, SVM, KNN, xgboost, and Adaboost algorithm	Androgens and estrogen , FSH , LH, AMH, TSH	High accuracy	99.99 %
Ahmad et al., 2024 [16]	Saudi Arabia	KNN, RF, multilayer perceptron, NB, SMOTE, CNN, LSTM	Testosterone and sex hormones	High accuracy	SMOTE + LSTM= 92.04% SMOTE + CNN = 96.59% SMOTE + CNN+ LSTM = 94.31%

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Author, Year	Country	Type of ML	Inputs Used	Other Outcomes	Accuracy
Lee et al., 2024 [17]	Finland and Sweden	CNNs	LH AMH FSH SHBG	The AI model had a low total error rate (6.32% for CD138– and 3.23% for CD138+), high agreement with human evaluations, and reduced intra/interobserver variations. No reported adverse effects.	95 %
Zhang et al., 2021 [18]	China	KNN, RF, XGB, and stacking models	GnRH FSH LH	No mention of adverse effects; false positives and negatives were evaluated via model performance metrics	89.32%
Alamoudi et al., 2023 [19]	Saudi Arabia	Deep Learning, CNNs, Transfer Learning, Fusion Models	Ultrasound images of ovaries, Clinical data (demographic, vital signs, laboratory tests, hormones)	False positives and false negatives were reported in the confusion matrices. The fusion model combining ultrasound images and clinical data outperformed models using only images or clinical data separately in terms of precision, F1-score, recall, and specificity.	Inception Model (Ultrasound Images Only): 84.81% accuracy Fusion Model (Ultrasound Images + Clinical Data): 82.46% accuracy
Mridul et al., 2024 [20]	Bangladesh	ANN, RNN, CNN, LSTM, BLSTM, Random FRF, LR, GB, KNN, ABC, DT, SVM, QDA), RC, PA, GNB	demographic data, clinical symptoms, hormonal profiles, imaging findings, and genetic marker	Some algorithms, such as KNN, exhibited a higher false positive rate (FPR)	98.27 %
Wu et al., 2023 [21]	China	ANN RF	10 genes (genetic biomarkers)	No adverse effects reported.	96.5 %
Nsugbe. 2023 [22]	UK	LSVM, QSVM, CSVM, FGSVM, MGSVM, CGSVM	Metabolic, physical, imaging, hormonal, and biochemical attributes from the Kaggle PCOS dataset. - medical health record	No mention of adverse effects; false positives and negatives were evaluated via model performance metrics	92 %
Dutta et al., 2021 [23]	India	Supervised Machine Learning- Logistic Regression, Random Forest, Decision Tree, Support Vector Machine, KNN	BMI, hormone: LH, FSH, DHEAS, Fasting insulin and Fasting blood sugar	False positives/negatives: Not explicitly mentioned. Adverse effects: Not applicable	97.11 %
M NS et al., 2024 [24]	India	Hybrid Machine Learning (SVM + Logistic Regression) with RMSprop optimization	17 attributes related to PCOS symptoms, including menstrual regularity, weight gain, hair growth, skin conditions, mental stress, and family history of diabetes and hypertension.	No Adverse Effects reported.	89.03 %
Neha et al., 2024 [25]	India	RF, LR, SVM, DT, NB, KNN, AdaBoost, XGBoost, Extra Trees,Ensemble Learning (Stacking Ensemble, Two-Level Random Forest Classifier), Explainable AI (XAI) techniques (SHAP, LIME,DALEX, PDP)	Numerical and categorical features related to PCOS, including hormone levels, lifestyle factors, and clinical parameters (e.g., follicle count, cycle length, BMI, weight, age, etc.)	The model achieved high accuracy (99.31%) with minimal false positives and negatives.The study highlights the importance of key features such as follicle count, cycle length, and BMI in PCOS prediction	99. 31 %
G UM et al., 2024 [26]		India	Supervised Machine Learning (Logistic Regression, SVM, NB, CART, RF)	Clinical data including age, weight, height, BMI, blood group, pulse rate, respiratory rate, hemoglobin levels, cycle length, marriage status, pregnancy history, hormone levels (FSH, LH, TSH, AMH, PRL, Vit D3, PRG), RBS, weight gain, hair growth, skin darkening, hair loss, pimples, fast food consumption, exercise habits, blood pressure, follicle count, and endometrial thickness.	The study did not report adverse effects or false positives explicitly, but it compared the performance metrics (accuracy, precision, recall, F1-score) of different algorithms.

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Author, Year	Country	Type of ML	Inputs Used	Other Outcomes	Accuracy
Rahman et al., 2024 [27]	Bangladesh		RF, AdaBoost, LR, DT, XGBoost, SVM, Multilayer Perceptron	Clinical data including age, weight, height, BMI, blood group, pulse rate, respiratory rate, hemoglobin levels, cycle length, marriage status, pregnancy history, hormone levels (FSH, LH, TSH, AMH, PRL, Vit D3, PRG), RBS, weight gain, hair growth, skin darkening, hair loss, pimples, fast food consumption, exercise habits, blood pressure, follicle count, and endometrial thickness.	No direct mention of adverse effects, but comparisons with other ML models show reduced false positives and improved detection rates
Khanna et al., 2023 [28]	India		LR, DT, RF, SVM, NB KNN AdaBoost XGBoost ExtraTrees Stacking models	Clinical parameters: BMI, pulse rate, blood pressure, menstrual cycle length, waist-hip ratio, Biochemical markers: LH, FSH, AMH, PRG, PRL, Hb, Beta-HCG Other attributes: Skin darkening, hair growth, weight gain, acne.	False positives and false negatives were analyzed using SHAP and LIME explainability tools. Balancing techniques like SMOTE were applied to mitigate class imbalance
Bharati et al., 2022 [29]	Bangladesh		Ensemble learning methods, including voting hard, voting soft, and CatBoost.	hormonal levels (FSH, LH, PRL, AMH, TSH, PRG), BMI, weight gain, cycle length, and follicle number.	False positives and false negatives are reported in the confusion matrix. The study highlights the reduction in computational time by using only 13 features.

Abbreviations: AMH: Anti-Müllerian Hormone, AND: Androstenedione, ANN: Artificial Neural Network, BLSTM: Bi-directional Long Short-Term Memory, BMI: Body Mass Index, BP: Blood Pressure, CART: Classification and Regression Tree, CNN: Convolutional Neural Network, DHEAS: Dehydroepiandrosterone Sulfate, DL: Deep Learning, DT: Decision Tree, FSH: Follicle-Stimulating Hormone, GAN: Generative Adversarial Network, GBM: Gradient Boosting Machine, KNN: K-Nearest Neighbors, LASSO: Least Absolute Shrinkage and Selection Operator, LH: Luteinizing Hormone, LR: Logistic Regression, LSTM: Long Short-Term Memory, MLP: Multilayer Perceptron, NB: Naïve Bayes, RF: Random Forest, RNN: Recurrent Neural Network, SGD: Stochastic Gradient Descent, SHBG: Sex Hormone Binding Globulin, SMOTE: Synthetic Minority Oversampling Technique, SVM: Support Vector Machine, TSH: Thyroid-Stimulating Hormone, UML: Upper Menstrual Length, USG: Ultrasonography, XAI: Explainable Artificial Intelligence, XGB: Extreme Gradient Boosting

accuracy rate of 85%- 98%. It is notable that SVM models were most effective when used with hormonal and metabolic markers such as AMH, LH/FSH ratio, and insulin resistance indices.

Even though these models are effective, researchers noted that for larger data sets they are a computationally expensive option and sensitive to feature scaling, requiring careful preprocessing [1,13].

The Random Forest model is an ensemble learning method that builds multiple decision trees and averages their outputs for better stability and accuracy. The studies analysed reported that this model is robust in handling missing data and feature redundancy with a high accuracy of 95%- 99% in PCOS datasets. As in other models, they are highly effective when trained on a combination of clinical, biochemical, and imaging features. It is noticeable that this model is computationally expensive and model interpretability is lower compared to Logistic Regression or Decision Trees [14,15,16].

KNN model, classifies new cases based on the majority class of k-nearest data points. Only a limited number of studies used this model and reported an accuracy ranging from 80%- 90%. KNN is effective for datasets with low noise and clear separation between PCOS and non-PCOS cases, but in larger datasets, their computational inefficiency will be a factor of concern, along with the requirement for careful tuning of k-values [4,14,17].

XGBoost (Extreme Gradient Boosting) is another important supervised learning model that uses gradient boosting techniques to create a strong classifier by iteratively correcting weak models. This model demonstrated high accuracy (96%-99%), often outperforming traditional supervised models. It is highly effective for structured clinical data and fast for larger datasets. Moreover, this model works well in imbalanced PCOS datasets by adjusting weighting factors. Although the complexity of training and tuning this model is high compared to other models, its accuracy and ability to handle larger datasets outweigh this challenge [1,18,19].

Unsupervised Learning models

In the unsupervised machine learning approach, the model finds hidden patterns in unlabeled data without predefined outcomes. In PCOS diagnosis supervised ML is dominant because labelled medical datasets are used as inputs, while unsupervised ML can help identify new PCOS subtypes. Examples of unsupervised MLs identified include, K-Means Clustering, Hierarchical Clustering, DBSCAN, Principal Component Analysis (PCA) etc (Figure 3).

The “K-Means Clustering” model groups data points into K clusters based on their similarity. The algorithm iteratively assigns each data point to the nearest cluster centroid until convergence. Studies used this model in PCOS metabolic profiling to classify patients into distinct metabolic-risk groups (e.g., insulin-resistant PCOS vs. normoinsulinemic PCOS) and also found that, PCOS patients

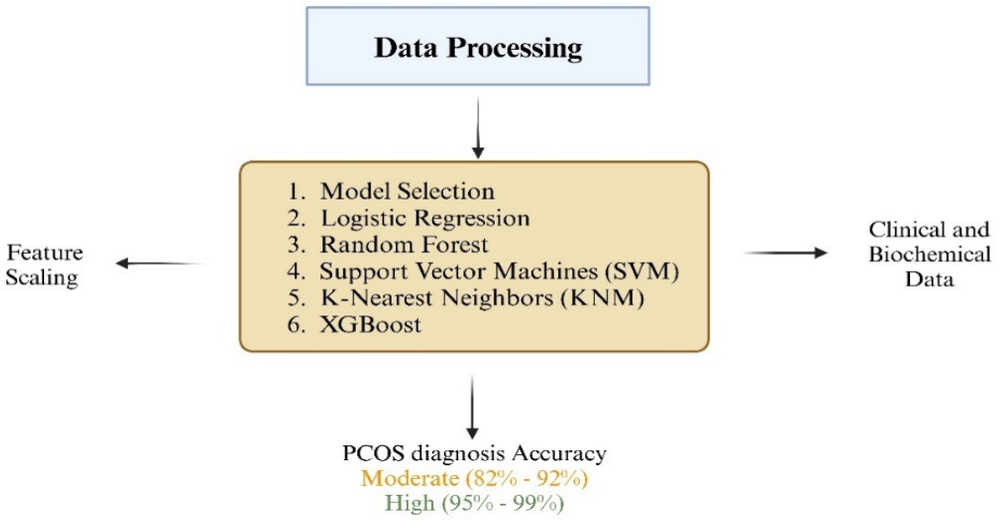

Fig. 2. Workflow of supervised machine learning models for the diagnosis of PCOS. Clinical and biochemical data undergo preprocessing, including feature scaling, followed by model selection and training using supervised algorithms such as logistic regression, random forest and SVM, KNN and XGBoost.

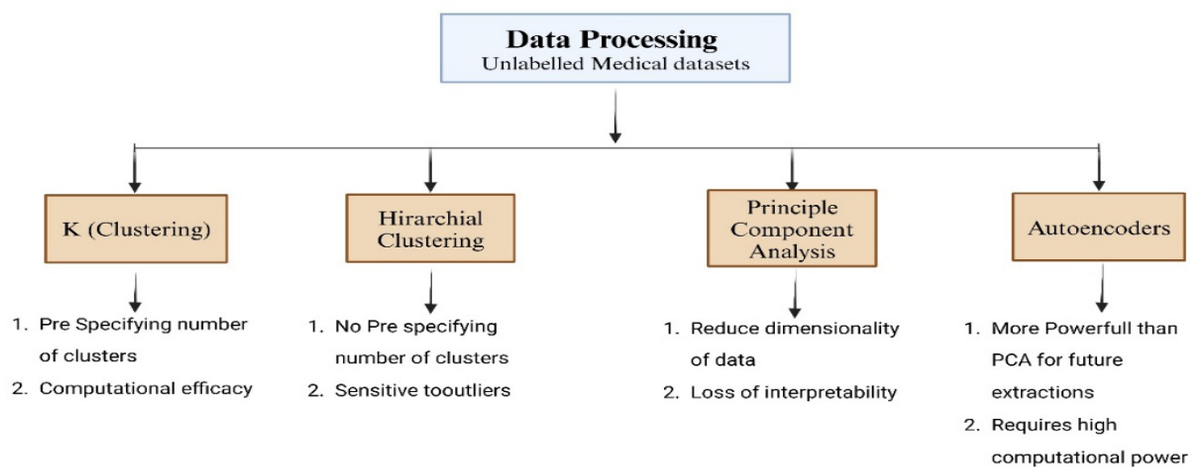

Fig. 3. Framework of unsupervised machine learning models applied for PCOS diagnosis using unlabelled medical datasets. The process begins with data preprocessing, followed by the application of various unsupervised learning techniques including K-means clustering (requiring pre-specification of cluster number and offering computational efficiency), hierarchical clustering (which is sensitive to outliers but does not require cluster pre-specification), principal component analysis (PCA) for dimensionality reduction at the cost of interpretability, and autoencoders, which are more powerful than PCA for feature extraction but demand high computational resources.

could be categorized into mild, moderate, and severe metabolic subtypes based on this. This is a computationally efficient model Effective for identifying phenotypic clusters within PCOS populations. However, this model requires pre-specifying the number of clusters (K), which may lead to incorrect grouping and does not handle overlapping PCOS subtypes well, as some cases have mixed metabolic and hormonal characteristics [20].

The hierarchical Clustering model builds a tree-like hierarchy of clusters, grouping data points based on their proximity. Studies used this model to identify subtypes of PCOS based on biochemical and clinical markers, for example classifying PCOS phenotypes based on hormonal vs. metabolic dominance. This model has some added advantage in that, it does not require pre-specifying the number of clusters, unlike K-Means and provides a detailed view of relationships between PCOS subtypes. Model is computationally expensive for large datasets and is Sensitive to outliers, which can distort clustering results [4,21,22].

The Principal Component Analysis (PCA) model reduces the dimensionality of data by transforming correlated variables into a smaller set of independent components, preserving variance. This model is applied by studies to identify key biomarkers driving PCOS classification and helped in removing redundant features, improving model efficiency. Even though this model can simplify high-dimensional PCOS datasets, it may lead to loss of interpretability as transformed variables do not have direct clinical meaning, moreover works best when features are linearly related, which may not always be the case in PCOS [23,24].

Autoencoders (Deep Learning-based Unsupervised Model) is a type of neural network that learns to compress and reconstruct input data, capturing key feature representations. Studies applied this model in PCOS prediction

models combining clinical and imaging data and was able to detect hidden correlations between PCOS markers. The added advantage of this model is that, it is more powerful than PCA for feature extraction and can be trained on large, unlabeled datasets. Even though, compared with traditional clustering methods, this model is not easy to interpret and requires high computational power [9,21,25].

Deep Learning models

Deep learning is a subset of ML that uses neural networks with multiple layers to automatically extract features and recognize patterns. Deep learning models like CNN (Convolutional Neural Networks), ResNet, YOLOv8 etc are used in identified studies. Among these CNN-based models, is highly effective in analyzing ultrasound images for PCOS diagnosis, extracting features such as follicle count and ovarian morphology with high accuracy [21,24,25] (Figure 4).

Convolutional Neural Networks (CNNs) are specifically designed to extract spatial features from image data. They utilize convolutional layers, pooling layers, and fully connected layers to identify patterns, making them particularly suitable for analyzing ultrasound images. Research studies have employed CNNs to assess ovarian ultrasound images, focusing on features such as follicle count, ovarian volume, and cystic patterns, with reported accuracies ranging from 95% to 99.89% [7,20]. A significant advantage of CNN models is their ability to detect subtle variations in ovarian morphology, alongside their capacity to automate feature extraction, which minimizes the need for human annotation. However, there are some limitations to consider, including the necessity for large, well-annotated image datasets for training and the high computational demands, which can increase the risk of overfitting when dealing with small datasets [7,26].

ResNet (Residual Neural Network) was applied in PCOS ultrasound classification tasks, often combined with other models such as U-Net for enhanced segmentation. One study reported over 92% accuracy using ResNet18 for follicle segmentation, showing robust feature extraction even in noisy medical images. ResNet is effective for deep architectures, enabling complex pattern learning without vanishing gradients. As in other deep learning models, ResNet also requires large-scale datasets and high computational resources, moreover, training time is longer compared to CNN [27].

Another important model is YOLOv8, which is an object detection framework optimized for real-time image analysis, capable of identifying multiple objects in a single frame. Studies used this model to detect ovarian cysts or follicles in ultrasound images of PCOS patients and combined with custom CNNs to improve classification accuracy to 97.5%, outperforming traditional detection techniques. YOLOv8 is a fast and accurate model suitable for real-time diagnostic tools. Just like in other models, YOLOv8 also requires custom fine-tuning for optimal performance in medical imaging [28].

Some studies combined deep learning for feature extraction with classical ML algorithms like Random Forest or XGBoost for final classification. A study integrated CNN-based image features with clinical data processed by XGBoost, achieving accuracy above 98%, whereas another study combined ResNet feature maps with an SVM classifier for better generalization. These hybrid deep learning models also require advanced integration pipelines and tuning.

Ensemble Learning

Ensemble Learning combines multiple ML models to improve accuracy and robustness by reducing biases and variance. Studies used Ensemble Learning models like Stacking models, Voting Classifiers, XGBoost, Bagging methods etc. The advantage of Ensemble Learning models over other models is, that they can enhance PCOS prediction accuracy by combining the strengths of Random Forest, Decision Trees, and Boosting methods, leading to more reliable diagnostic models [7,1,29] (Figure 5).

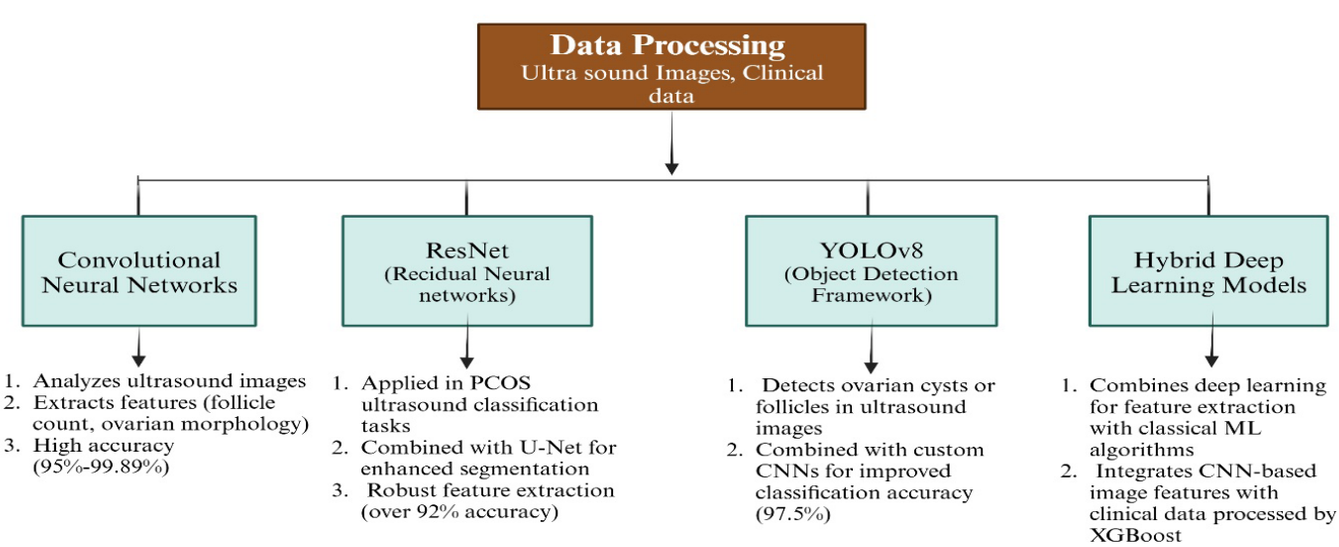

Fig. 4. Deep learning models identified for the diagnosis of Polycystic Ovary Syndrome (PCOS) using ultrasound imaging and clinical data.

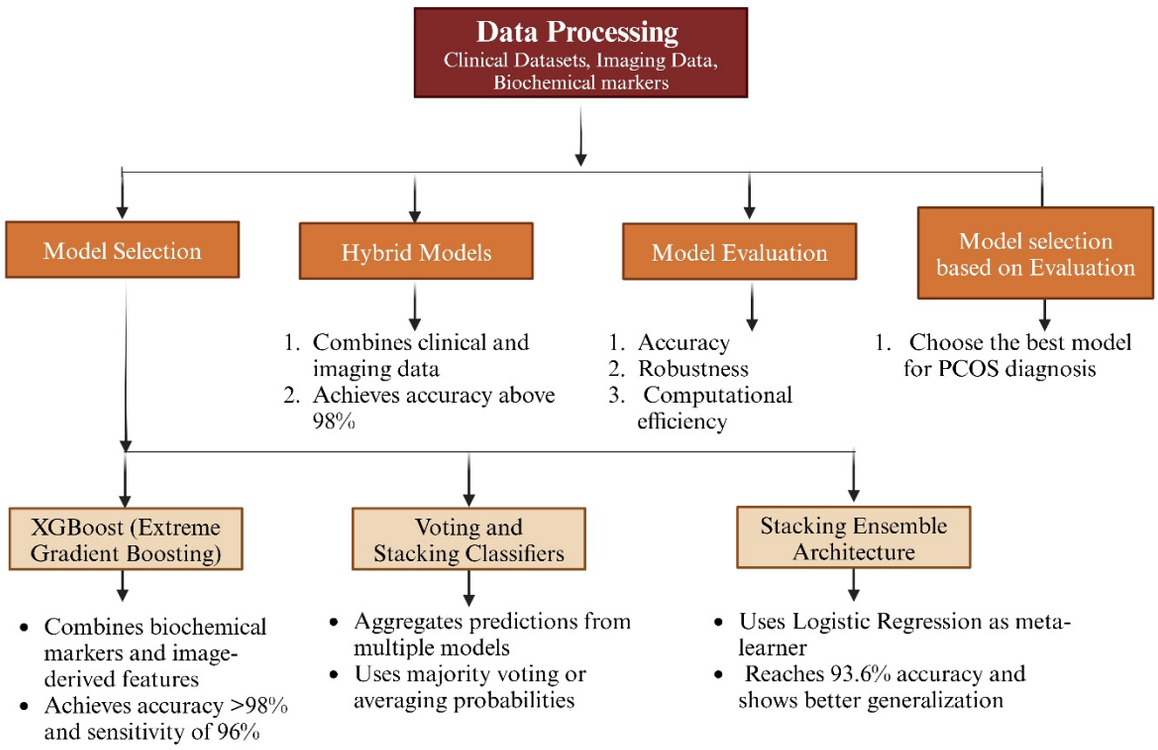

Fig. 5. Ensemble learning models identified for the diagnosis of Polycystic Ovary Syndrome (PCOS) using multimodal biomedical data

XGBoost (Extreme Gradient Boosting) is an optimized implementation of gradient boosting, which was widely adopted for its efficiency and scalability. XGBoost was used in a hybrid architecture combining biochemical markers and image-derived features. The model achieved accuracy >98% and sensitivity of 96%, outperforming both Random Forest and CNN when used independently [7,13]. Another study by Suha & Islam (2022) demonstrated XGBoost's superior performance in classifying PCOS using clinical datasets from South Asia. Here, ensemble boosting proved particularly valuable in handling imbalanced class distributions, which often skew predictions in PCOS research.

Another important ensemble learning model is the Voting and Stacking Classifiers. This model aggregate predictions from multiple models (e.g., SVM, KNN, Decision Trees) by majority voting (hard voting) or averaging probabilities (soft voting). Lv et al. (2022) applied a soft-voting ensemble combining SVM, Decision Trees, and Random Forest. The model achieved 95.2% accuracy, significantly higher than any of the base models alone. This fusion allowed the classifier to balance precision and recall, which is crucial for reducing false negatives in PCOS detection. Another study by Rachana et al. (2021) implemented a stacked ensemble architecture where Logistic Regression served as the meta-learner combining outputs from base models. This stacking ensemble reached 93.6% accuracy and showed better generalization on external validation datasets than individual models.

Almost all ensemble models reported accuracy above 90%, with some exceeding 98%, especially in hybrid models combining clinical and imaging data. However, models like XGBoost require intensive computation, especially in parameter tuning. The performance of ensemble models is often contingent on high-quality, well-pre-processed data. Poorly handled missing values or inconsistent labelling can reduce their effectiveness.

Hybrid Models

This is a combination of different ML approaches (e.g., Supervised + Deep Learning or Ensemble + Unsupervised Learning) to optimize performance. The best-performing combinations identified in the review are CNN + XGBoost, ResNet + Random Forest, Hybrid Neural Networks. Hybrid models are designed to combine clinical and imaging data, leveraging deep learning for image feature extraction and supervised ML for structured data analysis, offering a comprehensive diagnostic approach [15,23].

One of the most notable hybrid models was presented by Kermanshahchi et al. (2024), where a Convolutional Neural Network (CNN) was used to extract features from ovarian ultrasound images, and XGBoost was used as the final classifier. This combination resulted in an outstanding diagnostic accuracy of 98.9%. The CNN component learned spatial features such as follicle distribution, while XGBoost processed those features along with clinical inputs (e.g., BMI, AMH). This hybrid design enabled both high feature abstraction and classification robustness, addressing the shortcomings of using CNN or XGBoost alone.

In another hybrid approach, Rachana et al. (2021) implemented a model combining ResNet (a deep convolutional architecture) for feature extraction with a Support Vector Machine (SVM) as the classifier. This model was

particularly effective in distinguishing between PCOS and non-PCOS ovarian ultrasound images, reporting an accuracy of 94.2%.

Suha & Islam (2022) implemented a hybrid strategy that integrated CNN for image-based feature learning with an ensemble voting classifier (Random Forest + Logistic Regression + SVM) for diagnosis. This model achieved a classification accuracy of 95.6%. The hybrid ensemble component allowed the system to weigh decisions across models, reducing the likelihood of bias from any one classifier.

The hybrid ML models represent a significant advancement in the application of AI to PCOS diagnosis, where the studies have demonstrated that the hybrid architectures can outperform standalone models, achieving diagnostic accuracies exceeding 95%. These models are especially effective in multimodal analysis [7,19,29].

Input features used for ML model training

The effectiveness of an ML model largely depends on the quality and relevance of the input features used for training. Studies have used different input variables, ranging from hormonal markers (AMH, LH/FSH ratio, testosterone), metabolic indicators (BMI and insulin resistance), and clinical symptoms (hirsutism, menstrual irregularities), ultrasound imaging parameters (Ovarian volume, follicle count) (Figure 6). None of the included studies had a standardised approach to feature selection, which may have resulted in the variability in diagnostic performance [31]. Our qualitative analysis of the 34 studies indicates that no single feature can accurately diagnose PCOS. But a combination of biochemical, metabolic, and imaging features yields the best results.

Based on the comparative analysis, the most effective features for training ML models in PCOS diagnosis include; AMH and LH/FSH ratios, which are the strongest hormonal predictors of PCOS and ultrasound-based features are best for deep learning approaches. However, for higher accuracy and generalizability of results, a combination of different input features is preferable. These hybrid models integrating clinical, biochemical, and imaging data provide the most robust predictions, but require high computational power and large-scale datasets for optimal performance [30,31].

Discussion

The studies in our review consistently demonstrate that machine learning can markedly improve PCOS diagnosis accuracy, often exceeding 90% and even approaching near-perfect performance in controlled settings, especially deep learning models analyzing ultrasound images achieved exceptionally high accuracy. Suha and Islam (2022) reported 99.89% accuracy using a CNN with ensemble methods, and Kermanshahchi et al. (2024) even reported ~100% accuracy and sensitivity on their ultrasound image dataset. These results show accuracy of image-based deep learning to detect the polycystic ovarian morphology; however, they may also reflect overfitting or limited sample diversity, since perfect classification is rare in clinical practice [1, 11].

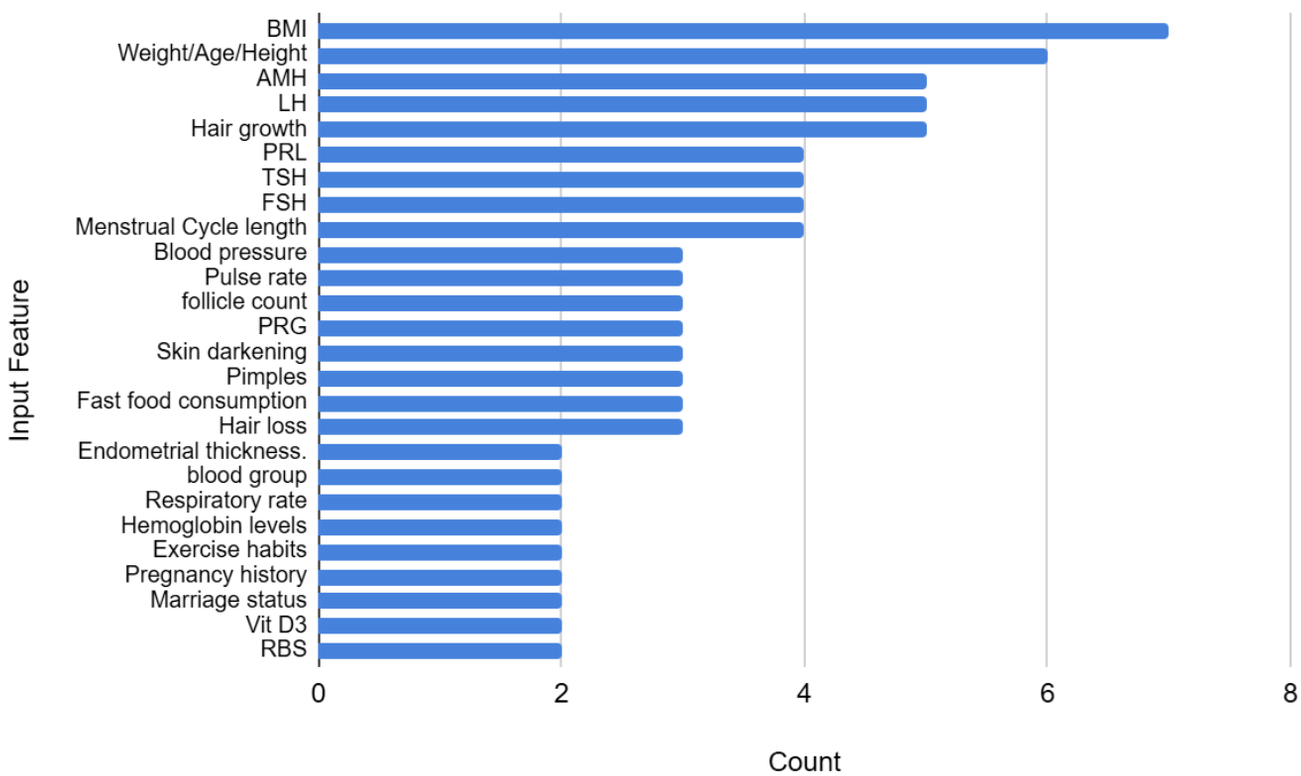

Fig. 6. Represents the most important input features used to train ML models. The bar graph illustrates how often these inputs were used in studies, with 70% of the studies incorporating BMI, while 50% included AMH, LH, and hair growth as key features. Only input features that were considered in at least 20% of the studies were included in the visualization [30,31].

In contrast, models using traditional clinical and biochemical data showed strong but slightly lower performance. For instance, Bharati et al. (2020) used a hybrid Random Forest-Logistic model on a clinical dataset and achieved about 91% accuracy with 90% recall. Similarly other studies found Random Forest most effective on tabular clinical features (attaining ~89–91% accuracy) [8,16, 29] . These conventional ML models benefit from interpretability and highlight key predictors (e.g. FSH/LH ratio, identified as highly informative by multiple studies), but they may miss complex nonlinear patterns compared to deep networks.

It is also notable that, not all approaches achieved such high accuracy, a few demonstrated the challenges in PCOS detection.Sumathi et al. (2021) developed a CNN for classifying ovarian cysts and reached only ~85% accuracy, likely due to limited training data and a simpler network. Likewise, Lim et al. (2023) explored a novel non-invasive diagnostic avenue using radial pulse wave analysis, but reported a modest overall accuracy of 72%. This lower performance suggests that emerging modalities (e.g. pulse signals or experimental biomarkers) may not yet be as sensitive or specific as conventional hormone and imaging features [6,14].

Some models in the literature emphasize sensitivity, whereas others sacrifice sensitivity for specificity [11]. Luo et al. (2019)'s logistic model using three serum biomarkers attained 82.1% sensitivity at 92.3% specificity, a respectable balance, but it could miss nearly 18% of cases [5]. In summary, while many ML models show excellent diagnostic accuracy for PCOS, the few that underperform highlight that adequate data quality, feature selection, and model choice are crucial to achieving both high sensitivity *and* specificity. Models built on small or narrowly selected populations (e.g. non-obese women) or unconventional data sources tend to have lower generalizability.

The ensemble and hybrid models have emerged as top performers in several studies, Alam Suha et al. (2023) combined five classifiers in a stacking ensemble and improved accuracy to ~95.7%, notably reducing false positives/negatives compared to any single algorithm [1]. Ensemble methods capitalize on the complementary strengths of different algorithms, often yielding higher robustness. However, they can be complex and less interpretable. Simpler classifiers like Support Vector Machines and Random Forests were frequently among the best performers in individual [25]. These supervised learning models are relatively interpretable and, with proper feature selection, can achieve high accuracy with lower computational cost.

On the other hand, deep learning particularly CNNs, excels in tasks involving imaging data. Multiple studies showed that CNN-based analysis of ovarian ultrasound images can detect polycystic morphology with high precision; Rachana et al., 2021 achieved 97% accuracy using a KNN classifier after image feature extraction, and a custom CNN "F-Net" by Sowmiya et al., 2024 achieved ~97.5% on ultrasound images [3,4]. Deep networks can capture subtle features, such as follicle distribution or stromal texture beyond human vision, but they require larger datasets for training; when sample size was small, performance could suffer or models risk overfitting [11].

Across these diverse approaches, certain predictors of PCOS consistently emerged. Features related to reproductive hormones were dominant: the LH to FSH ratio was highlighted as a pivotal feature in numerous studies, aligning with clinical understanding of PCOS endocrine profiles. High serum AMH levels, reflecting increased follicle count, also frequently contributed to model predictions [10]. Anthropometric and metabolic features like BMI, ovarian ultrasound follicle count, menstrual cycle irregularity, and markers of insulin resistance are the other important inputs in many models. These common predictors underscore that ML algorithms, when properly trained, are identifying known hallmarks of PCOS.

Nonetheless, some models have incorporated novel features also, such as Lv et al. (2022) successfully used ocular images (scleral blood vessel patterns) to detect PCOS with ~93% accuracy, hinting at systemic manifestations of PCOS that could expand diagnostic modalities [2]. Likewise, a few studies have explored genetic and molecular markers as inputs, which may improve understanding of PCOS pathophysiology. These novel approaches, while promising, often suffered from small sample sizes and require further validation.

This collective evidence suggests that ML, if rigorously developed, could become a valuable adjunct to conventional PCOS diagnosis. Currently, the diagnostic process for PCOS is fraught with variability, different specialists may emphasize Rotterdam criteria vs. androgen excess criteria, leading to inconsistent diagnoses. AI models offer a chance to standardize this by objectively combining multimodal data.

Another emerging benefit of ML in this field is the ability to uncover PCOS subtypes and comorbid risk patterns. Since PCOS is heterogeneous in clinical presentation, clustering and advanced models could stratify patients into phenotypes, each requiring tailored management [31].

Despite these promising results, our systematic review is not without limitations. Our synthesis depended on majority of studies that relied on single-center datasets with limited ethnic diversity, restricting external generalizability. Very few studies validated models using independent external cohorts, raising concerns about overfitting. Moreover, the lack of standardized feature selection frameworks contributed to wide variability in input data, complicating direct comparison between models. In addition to all, we restricted inclusion of our review to English-language and open-access publications from 2014–2024, which may have excluded relevant studies published outside these criteria.

There is an inherent publication bias toward positive findings in this field, studies reporting high accuracies are more likely to be published, skewing the overall picture.

We also observed high heterogeneity in study designs and outcome reporting. A formal meta-analysis was not feasible due to differing performance metrics such as accuracy, AUC, F1-score and varied endpoints where, some focused on binary diagnosis, others on subclassification. Instead, we synthesized the findings qualitatively.

Despite these caveats, the insights gained are valuable for guiding future research. Our review identifies successful strategies that future studies can build upon. It also flags common pitfalls to avoid, such as lacking external validation or ignoring the interpretability of models. Moving forward, larger multi-center studies and prospective trials are needed to test the most promising ML models in real clinical workflows. Collaboration between data scientists and clinicians will be key to ensure that models address real-world diagnostic challenges and are evaluated against current gold standards. In summary, even with certain limitations, this review provides a comprehensive overview of the state of ML in PCOS diagnosis and underscores a clear trend: data-driven approaches hold great promise to augment clinical decision making in PCOS, offering earlier detection and more consistent, personalized care. Future studies can leverage our findings to refine AI models aiming not just for high accuracy in a research setting, but for robust, generalizable tools that improve patient outcomes in everyday clinical practice.

Conclusion

Machine learning models have significant potential to improve diagnostic accuracy in the identification of Polycystic Ovary Syndrome. Supervised models have shown high predictive accuracy when trained on structured clinical and biochemical datasets. Deep learning models have proven effective in analyzing ultrasound images for automatic feature extraction. Moreover, hybrid and ensemble approaches have surpassed individual models by combining multiple data types, which enhances their robustness and generalizability. The results of this systematic review prove that the integration of AI-driven diagnostic tools offers great promise for achieving personalized, timely, and accurate diagnoses of PCOS, potentially transforming women's reproductive healthcare.

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Authors' contributions

AVP (Conceptualization; Data curation; Formal analysis; Investigation; Methodology; Project administration; Resources; Software; Supervision; Validation; Visualization; Writing – original draft; Writing – review & editing)
RS (Conceptualization; Formal analysis; Investigation; Methodology; Supervision; Writing – original draft; Writing – review & editing)
K (Data curation; Formal analysis; Methodology; Resources; Validation; Supervision; Writing – review & editing)
DNS (Data curation; Investigation; Methodology; Visualization; Writing – original draft; Writing – review & editing)
PV (Investigation; Resources; Validation; Writing – review & editing)
BV (Data curation; Formal analysis; Visualization; Writing – review & editing)
MT (Investigation; Methodology; Resources; Writing – review & editing)
SM (Investigation; Validation; Visualization; Writing – review & editing)
SK (Conceptualization; Supervision; Writing – review & editing)
LSJ (Resources; Supervision; Validation; Writing – review & editing)
BB (Formal analysis; Validation; Writing – review & editing)
MA (Software; Resources; Writing – review & editing)
SS (Visualization; Methodology; Writing – review & editing)
LS (Data curation; Resources; Validation; Writing – review & editing)

Conflict of interest

None to declare.

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