Advancing Polycystic Ovary Syndrome Detection with Artificial Intelligence Techniques

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Abstract: Polycystic Ovary Syndrome (PCOS) is a common hormonal disorder that affects women of reproductive age. Diagnosis mainly relies on traditional methods, such as clinical evaluations or laboratory tests, which can be expensive and time-consuming and are often accompanied by complex imaging techniques. The integration of Artificial Intelligence (AI), namely Machine Learning (ML) and Deep Learning (DL), seems to offer promising opportunities, allowing for the analysis of large datasets to improve PCOS detection and management. This work conducts a systematic literature review and aims to explore how ML and DL can optimize PCOS diagnosis by analyzing the most used data and algorithms while following a rigorous methodology to ensure the validity of the results. It also discusses the explainability of AI methods to be used by healthcare professionals, who are always looking for reliable results to support the best possible diagnosis for their patients.

1 INTRODUCTION

PCOS is a common hormonal disorder affecting women of reproductive age. It is characterized by a range of symptoms such as irregular menstrual cycles, hyperandrogenism (excess male hormones), the presence of multiple ovarian cysts, significant hair loss, and notable weight gain (Narinder et al., 2023). PCOS is often associated with ovarian dysfunctions that can lead to miscarriages, infertility, and even gynecological cancers. The syndrome has also a significant financial and psychological impact on patients. Thus, it becomes evident that the improvement of early detection tools for PCOS are crucial (Dana et al., 2022).

Given these major challenges for women with PCOS and the impact on their daily lives, Artificial Intelligence (AI) seem to offer promising prospects for improving the understanding, diagnosis, and management of PCOS (NMerlin and Sangeetha, 2023). Several works have addressed this emerging research issue (Ajil et al., 2023; Srivastav et al., 2024). Adopting AI for PCOS diagnosis seems promising but some challenges must be addressed for a secure and successful application.

In this work, we propose a Systematic Literature Review (SLR) to answer the following research issue: How is AI presenting new perspectives for detecting PCOS? Our article presents several contributions: We adopted a rigorous research methodology to conduct our SLR regarding this issue. We analyze the selected papers deeply to handle 3 main aspects: The features used in PCOS detection with AI, the ML, and DL models showing the best performances and the consideration of explainability in existing PCOS detection works. We also give a critical eye to existing works, highlighting the main challenges that can give insights into future works.

The remainder of this paper is organized as follows: We first provide background information about PCOS diagnosis and the importance of using AI to detect it, and previous literature reviews in Section 2, before detailing our methodology for this SLR in Section 3. In Section 4, we present the analysis and the results of our SLR and then discuss the challenges associated with this research question in Section 5. Finally, we draw our conclusions, with insights in future works in Section 6.

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2 BACKGROUND AND PREVIOUS LITERATURE REVIEWS

In this section, we present the background of the subject and review the literature reviews that have addressed this topic in previous years.

2.1 Polycystic Ovary Syndrome

PCOS is the most common endocrine disorder among women of reproductive age, affecting 10% to 15% (Narinder et al., 2023), and, according to INSERM (the French National Institute of Health and Medical Research), is the leading cause of female infertility. PCOS can be identified through imaging as an abnormal enlargement of the ovaries (>10 ml in volume) or many small follicles (>20 follicles smaller than 9 mm in diameter), as shown in Figure 1.

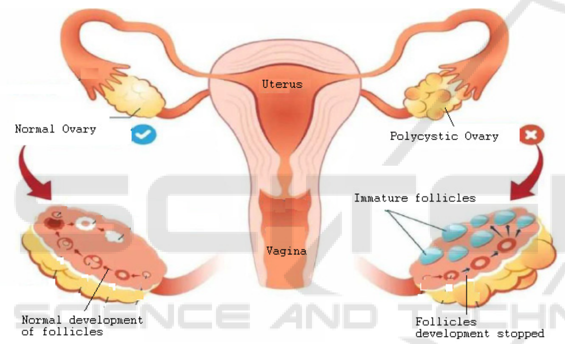

Figure 1: Polycystic Ovary Syndrome (PCOS) (Narinder et al., 2023).

PCOS is characterized by the ovaries producing an abnormal number of androgens, male sex hormones that are normally present in small quantities in women. This overproduction of hormones can lead to various complications, including anovulation, causing infertility, diabetes and irregular menstrual cycles, along with fatigue, anxiety, or depression (Ajil et al., 2023).

- **PCOS Detection**

The diagnosis of PCOS is usually made by gynecologists or midwives, who follow a step-by-step process. The difficulty in detecting PCOS arises from the fact that most symptoms can easily be misattributed to puberty or stress. This is why blood tests and laboratory analysis must be added to this procedure to obtain results on hormone levels in the blood. However, according to the midwives interviewed, INSERM, and (NMerlin and Sangeetha, 2023), these results must also be combined with imaging, such as MRI or pelvic and/or transvaginal ultrasounds.

- **Using AI to Detect PCOS:**

The use of AI for the detection and management of PCOS represents a major challenge in the medical field. AI can accurately count the number of cysts present in the ovaries (with a threshold of at least 20 immature follicles, each measuring less than 9 mm in diameter, according to INSERM). Thanks to its ability to analyze large datasets, AI enables the faster and more precise identification of PCOS clinical signs, thereby reducing diagnostic delays. However, this advancement raises significant challenges, particularly regarding the interpretability of results by healthcare professionals.

2.2 Previous Literature Reviews

Other works studied the existing literature on the use of AI for PCOS detection: (Barrera et al., 2023) compared the AI algorithms used in the literature and expressed reservations about their results due to the heterogeneity of the available data and the risk of bias. In general, ethical aspects and the explainability of the algorithms were not addressed, although a willingness to collaborate with the medical community during validation tests is mentioned. In their SLR, (Suha and Islam, 2023) mainly focused on the methodology, results, recommendations, and technical challenges related to the diagnosis of PCOS. Even though they mentioned explainable AI, (Suha and Islam, 2023) did not provide examples of application in the context of PCOS, making it difficult to assess the relevance of the proposed methods and their potential impact on medical practice, which limits the practical scope of the presented research. Moreover, (Graselin et al., 2023) focused on ML approaches for the detection of PCOS by evaluating the effectiveness, techniques, and results of previous studies; highlighting their technical shortcomings.

Besides, (Ahmed et al., 2023) provided an in-depth analysis of various ML and DL approaches for diagnosing PCOS. However, it does not address the transparency or reproducibility of the research. Ethical aspects and algorithm explainability were not addressed.

In our work, we followed a rigorous research protocol and carefully collected and analyzed research articles to conduct an SLR dedicated to the use of AI for PCOS detection. What distinguishes our work from others is that we deeply focus on each aspect of the PCOS detection process using AI. We start by identifying the most common data and features for the diagnosis of PCOS. We then highlight the algorithms that have shown the best performance. We investigate the explainability of AI techniques by healthcare practitioners in the context of PCOS. We emphasize the implications that could govern the use of AI for PCOS diagnosis and give insight into future work.

3 RESEARCH METHODOLOGY

An SLR is a rigorous research method that synthesizes evidence from the literature (Petersen and Vakkalanka, 2015; Keele, 2007). In the context of PCOS detection, this approach provides a comprehensive and objective view of current diagnostic methods while gathering and synthesizing evidence from the literature.

Research Questions:
We have structured our initial research issue into 3 Research Questions (RQ):

- **RQ1:** What are the most used features for diagnosing PCOS using AI?
- **RQ2:** Which machine learning/deep learning algorithms have shown the best performance?
- **RQ3:** Is explainability considered to increase trust in diagnostic models?

Search Query:
The following search query was executed on the Scopus library:
(detection OR diagnostic OR diagnosis) AND (PCOS OR "Polycystic ovary syndrome" OR "Polycystic Ovarian Syndrome") AND ("Machine Learning" OR ML OR "Deep Learning" OR DL).

To focus on recent works, we selected papers published from 2017 to 2024.

Papers Selection:
To summarize the synthesis of the scientific literature, we use the PRISMA model (Preferred Reporting Items for Systematic Reviews and Meta-Analyses) (Rethlefsen and Page, 2022).

Our research query returned initially n=43 articles. We applied inclusion and exclusion criteria.

Our Inclusion criteria are: (a) Paper addressing PCOS through machine learning, (b) Paper published from 2017 to 2024 (to focus on recent studies), and (c) Paper accessible online. Our exclusion criteria are: (a) Papers not written in English, and (b) Papers not peer-reviewed (such as abstracts, poster, proposal, technical reports, thesis). We had to exclude 20 articles for reasons of inaccessibility, 2 systematic reviews, and 1 paper not written in English. We obtained 20 papers after this first selection. The snowballing method allowed us to integrate two new articles into our corpus, bringing the total number of articles to 22.

4 ANALYSIS

This section is dedicated to SLR results, presenting responses to our three research questions.

Table 1: Overview of datasets used in studies for diagnosing PCOS with ML.

Dataset name	Type of data	Details	Ref.
PCOS (kottarathil, 2018)	clinical, metabolic, physical, hormonal, ultrasound imaging data, lifestyle, and social factors.	541 women (177 with PCOS, 364 without). 43 features.	(Ajil et al., 2023 ; Denny et al., 2019 ; Modi and Kumar, 2024 ; Subha et al., 2024 ; (Khanna et al., 2023 ; (Batra et al., 2023 ; Tanwar et al., 2023)
PCOS detection using ultrasound images (Choudhari, 2020)	Ultrasound imaging	3856 ultrasound images of women aged 22 to 39. Images are classified as infected or non-infected.	(Diptho et al., 2023; NMerlin and Sangeetha, 2023; Rashid et al., 2023; Prasher and Nelson, 2023 ; Hosain et al., 2022 ; Srivastav et al., 2024 ; Narinder et al., 2023)
Polycystic Ovary Ultrasound Images Dataset (Adiwijaya and Astuti, 2021)	Ultrasound imaging	54 ultrasound images from 14 PCOS patients and 40 controls. Images were captured using a vertical probe ultrasound device.	(G¨ulhan et al., 2023)
Ovarian ultrasound image (Dewi et al., 2020)	Ultrasound imaging	Contains ultrasound images validated by a gynecologist, including both infected and non-infected ovaries.	(Dewi et al., 2020)

4.1 What Are the Most Used Features for Diagnosing PCOS Using AI (RQ1)?

To answer this RQ, we analyzed the datasets used for training and testing ML algorithms. We then examined the different types of data used in these models. We studied the feature selection methods employed and conducted an in-depth analysis to determine the features most frequently selected and used by AI algorithms for PCOS diagnosis.

4.1.1 Dataset Analysis

We identified four datasets that we describe in Table1.

4.1.2 Data Type

Based on Table 1, datasets 'PCOS' and 'PCOS detection using ultrasound images are the most frequently used in the literature. The first dataset integrates clinical, metabolic, physical, hormonal, and ultrasound imaging data and data related to lifestyle and social factors. In contrast, the second dataset focuses exclusively on medical imaging.

To better in-depth analysis and understanding of the various data involved in the diagnosis and management of PCOS, we organized the data into three distinct categories: clinical data, non-clinical data, and medical imaging data.

- **Clinical Data:**

Related to a patient's health collected during medical care or clinical research. It can come from various sources and is used by healthcare professionals to make informed decisions regarding patient care. It can include for example blood type, Hemoglobin level, levels of vitamin D3, heart rate per minute, respiratory rate per minute, cycle duration, etc.

- **Non-Clinical Data:**

Can be derived from different aspects of lifestyle, such as regular exercise or frequent fast-food consumption. It may also include social factors, such as the duration of the patient's marriage. It also concerns physical features such as age, weight, height, body mass index, etc.

- **Medical Imaging Data:**

Represents ultrasounds used to visualize the ovaries and identify the typical cysts associated with PCOS.

4.1.3 Most Selected Features

Feature selection is a crucial step in ensuring the performance of a ML model. It involves identifying the most relevant variables that can effectively distinguish whether a patient has PCOS or not. Various statistical methods are used to select these relevant features such as ANOVA (Analysis of Variance), Pearson and Spearman Correlations and Chi-Square Test. Among the non-clinical features, 'Hair growth' stands out as the most important variable, followed by 'weight gain', 'skin darkening' and 'hair loss'. The presence of 'fast food' consumption is also noted as a significant factor. These symptoms are crucial for the ML model to effectively diagnose the disease.

In terms of clinical features, the most prominent features include '*left follicle size*', '*right follicle size*', '*normal follicles on the left*', and whether '*the cycle is regular or irregular*'.

4.2 Which AI Algorithms Have Shown the Best Performance (RQ2)?

To detect PCOS, several AI models were used. We differentiate these works based on the data (features) used. Table 2 presents an overview of AI models used for PCOS detection according to each data type.

Data preprocessing is crucial for optimizing a model's overall performance. It ensures data quality, making sure that the features used in ML models are reliable and accurate. Additionally, it preserves data integrity by consistently normalizing and scaling the inputs (Tanwar et al., 2023). The preprocessing of clinical and non-clinical data involves cleaning and transforming the data to make it usable by the model. It ensures data quality and integrity, allowing the ML model to function optimally and produce more accurate and reliable results. It helps correct errors and/or remove elements that could distort the results (NMerlin and Sangeetha, 2023; Tanwar et al., 2023). Researchers process as well by **dimensionality reduction**, helping to overcome the challenges posed by an excess of features while retaining as much information as possible (Denny et al., 2019). As for **data transformation,** it enables the conversion of categorical data into a numerical format that can be used by ML algorithms, thereby ensuring fair treatment of all variables (Batra et al., 2023).

After analyzing all these papers, we summarize the results of the Best Performances of AI models in terms of accuracy in Figure 2. For clinical and Non-clinical Data, CatBoost (Modi and Kumar, 2024) present the best accuracy. Concerning Images, the best accuracy is given by AMCNN (Rashid et al., 2023), Inception V3 (Narinder et al., 2023), and CNN/ VGG16 Model (Srivastav et al., 2024], while the INCEPTION Model and Light GBM in (NMerlin and Sangeetha, 2023) give the best performance for mixed data.

Table 2: Overview of AI models used for PCOS detection according to each data type.

Data type	Used algorithms	References
Clinical/ Non clinical	KNN/ NN/ NB/ SVM/ ClassificationTree (CT)/ LR	(Dana et al., 2022)
	AdaBoost/ XGBoost/ DT/ RF/ LR	(Ajil et al., 2023)
	LR/ KNN/ NB/ SVM	(Denny et al., 2019)
	CatBoost/ LR/ RF/ KNN/ NB/ SVM/ AdaBoost	(Modi and Kumar, 2024)
	RF/ XGB	(Subha et al., 2024)
	SHAP/ DNN	(Khanna et al., 2023)
	RF/ SVM/ LR	(Batra et al., 2023)
	RF	(Tanwar et al., 2023)
	LR/ RF/ NB /KNN	(Rao et al., 2024)
	AdaBoost/ NB/ DT/ KNN/ SVM/ XGBoost/ RF	(Syed et al., 2023)
Images	Inception V3	(Narinder et al., 2023)
	AMCNN	(Rashid et al., 2023)
	LSTM/ BI-LSTM	(Diptho et al., 2023)
	CNN	(Prasher and Nelson, 2023; Srinithi and Rekha, 2023; Srivastav and Krishnamoorthy, 2023; Dewi et al., 2020)
	PCONet/ Inception V3	[(Hosain et al., 2022)
	VGG16 Model	(NMerlin and Sangeetha, 2023)
	Squeeze Net/ CNN	(G¨ulhan et al., 2023
Mixed data	Inception Model and Light GBM	(NMerlin and Sangeetha, 2023)

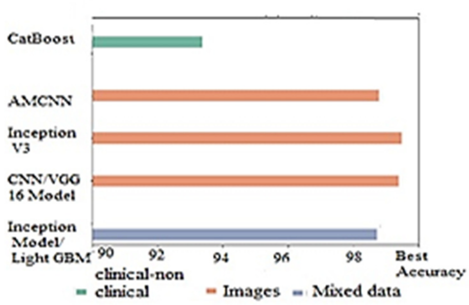

Figure 2: Best Model Accuracy by Data Type.

4.3 Is Explainability Considered to Increase Trust in Diagnostic Models (RQ3)?

Most works on ML in the PCOS context focus on algorithm performance, considered the main criterion for comparing different methods and a key measure of their effectiveness. This emphasis on metrics like '*accuracy*', '*recall*', or other performance measures often means that many studies neglect the transparency of results and the explainability of model decisions.

Certain ML algorithms are referred to as 'white-box', such as LR and DT; while others are referred to as 'black-box', such as Deep Neural Networks, RF, XGBoost and Adaboost. To address the limitations of *black-box* models, Explainable Artificial Intelligence (XAI) methods have been developed (Gade and Taly, 2019). In Table 3, we detail each study's use of white-box methods as well as the black-box algorithms employed. We also mention the name of the XAI method, if it exists.

Table 3 reveals that only one study has addressed the transparency of black-box models by using XAI techniques. This study stands out for integrating LIME (Local Interpretable Model-agnostic Explanations) and SHAP (SHapley Additive exPlanations), which provide interpretability to complex models. We can also see that several methods rely on white-box algorithms, even though these algorithms are not the most effective. Other studies focus solely on performance metrics without examining interpretability, highlighting a gap in research regarding model transparency, particularly in critical areas such as patient care and medical diagnosis.

Table 3: Use of white-box, black-box, and XAI methods.

White-box methods	Black-box methods	XAI method(s)	Ref.
Linear Discriminant Classifier (LDT), LR, Classification Tree (CT), KNN, SVM, NB	NN	–	(Dana et al., 2022)
LR, KNN, SVM, NB, DT	AdaBoost, XGBoost, RF	–	(Ajil et al., 2023)
LR, Classification and Regression Tree (CART), KNN, SVM, NB	RF	–	(Denny et al., 2019)
LR, KNN, SVM, NB	CatBoost, RF, AdaBoost	–	(Modi and Kumar, 2024)
SVM	RF, XGB	–	(Subha et al., 2024)
–	DNN	SHAP LIME	(Khanna et al., 2023)
LR, KNN, SVM, NB	RF	–	(Batra et al., 2023)
NB	RF	–	(Tanwar et al., 2023)
LR, KNN, SVM, NB	RF	–	(Rao et al., 2024)
NB, KNN, SVM, DT	AdaBoost, RF	–	(Syed et al., 2023
–	Inception V3	–	(Narinder et al., 2023)
–	CNN, LSTM, BI-LSTM	–	(Diptho et al., 2023)
–	AMCNN (Attention-based Multi-Channel Neural Network)	–	(Rashid et al., 2023)
–	CNN	–	(Prasher and Nelson, 2023; Dewi et al., 2020; Srinithi and Rekha, 2023; Srivastav and Krishnamoorthy, 2023)
–	PCONet (Pose-Conditioned Network), Inception V3	–	(Hosain et al., 2022)
–	CNN/VGG16	–	(Srivastav et al., 2024)
–	Squeeze Net/CNN	–	(G¨ulhan et al., 2023)
–	Inception Model and Light GBM	–	NMerlin and Sangeetha, 2023)

5 DISCUSSION

By addressing our three research questions, we identified a set of challenges that remain to be tackled, as well as research gaps to explore. The first challenge concerns the datasets used in the studies. There is an urgent need to create mixed datasets that incorporate both clinical data, non-clinical data, and medical images. Data diversity is crucial for improving the robustness and generalization of diagnostic models. Currently, many studies focus on isolated data, which may limit the models' ability to capture all aspects of PCOS. In the same context, why not rely on mixed features that combine different types of data? By integrating variables from various sources, such as biological analyses, medical histories, and physiological characteristics, we could improve diagnostic accuracy. The second challenge concerns the explainability of the models. While ML and DL algorithms can provide impressive performance, it is equally important to ensure that these models are interpretable and transparent. Healthcare practitioners must be able to understand the reasoning behind the models' predictions to establish trust and ensure that clinical decisions are based on solid foundations.

The lack of integrated datasets, the absence of a multi-feature approach, and the neglect of explainability are remaining reseach gaps, highlighting important opportunities for future research. Furthermore, the integration of AI in the detection of PCOS raises critical issues related to ethics and the regulation of health data. There are best practice guidelines that allow the use of sensitive data, such as health data, within an ethical framework. These universal rules include, among others: consent, transparency, bias prevention, anonymization, and peer validation of a model. The consideration of ethical concerns represents a major research gap in PCOS detection that must be considered in future works.

6 CONCLUSION AND FUTURE WORKS

In this paper, we performed an SLR, describing how AI, can present new perspectives for PCOS detection; highlighting three aspects: Features used for PCOS detection with AI; AI algorithms showing the best performance; and if explainability is considered to increase trust in diagnostic models. The advances enabled by AI in PCOS detection are promising.

However, there still are many challenges: The lack of integrated datasets, combining clinical, non-clinical data and images; the absence of a multi-feature approach and the lack of focus on explainability that underscores significant opportunities for future research. Thus, we aim to propose an enhanced PCOS detection system, addressing the limitations of existing works by integrating multi feature data, focusing on XAI methods, to provide Healthcare practitioners with interpretable results.

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