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Phenomapping Polycystic Ovary Disorder Using Self Supervised Representation Learning and Deep Clustering of Clinical Data

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Abstract: Poly cystic Ovary Disorder is one of the most prevalent endocrine conditions affecting women of reproductive age, yet its clinical presentation varies widely across reproductive metabolic and hormonal domains. Current diagnostic frameworks and computational models often simplify PCOD into a binary classification present or absent ignoring its intrinsic heterogeneity. This oversimplification hinders precision medicine and the development of tailored treatment plans. This paper introduces a novel framework that combines self-supervised learning (SSL) and deep clustering to uncover hidden PCOD phenotype directly from unlabeled clinical records. Contrastive SSL approaches such as Sim CLR and MoCo are used to learn patient embeddings which are subsequently clustered using Deep Cluster and SwAV. The resulting clusters are validated against clinical indicators including body mass index (BMI) hormonal ratios and insulin resistance measures. Experiments revealed three distinct clusters: mild PCOD with near normal parameters moderate PCOD with hormonal irregularities and severe PCOD with metabolic risks. The framework was deployed in a Django based clinical platform providing real time cluster assignments visual analytics and patient level reports. By moving beyond binary classification this work demonstrates the potential of SSL driven phenomapping to enable precision gynecology supporting individualized treatment strategies and advancing scalable clinical decision support systems.

Keywords: Polycystic Ovary Disorder, self-supervised learning, contrastive learning deep clustering, clinical phenotyping, decision support.

I. INTRODUCTION

Poly cystic Ovary Disorder (PCOD) also known as Poly cystic Ovary Syndrome (PCOS) affects between 8% and 12% of women globally during their reproductive years. Its symptoms range from irregular ovulation and infertility to metabolic disturbances such as insulin resistance obesity and an increased risk of type 2 diabetes and cardiovascular disease. This variability highlights PCOD not only as a reproductive disorder but also as a systemic condition.

Diagnostic standards such as the Rotterdam criteria and NIH definitions provide a foundation but fail to capture the full diversity of PCOD phenotype. As a result, many women remain diagnosed or receive generic treatment plans. For instance, lean PCOD patients and obese PCOD patients may exhibit vastly different prognoses and require different interventions. Machine learning (ML) has shown promise in PCOD analysis, but existing models are largely supervised and treat the condition as binary. This restricts their ability to reveal nuanced sub types Moreover supervised learning requires large, annotated datasets which are often scarce in clinical practice. To address these limitations, we propose a data driven unsupervised framework that combines self-supervised learning and deep clustering to identify hidden PCOD phenotype. The approach is implemented in a clinician friendly Django platform that provides real time predictions and personalized reports. First application of contrastive SSL for PCOD sub type discovery. Integration of Deep Cluster and SwAV for unsupervised phenotype identification. Development of a clinical decision support system for real time patient reporting. Poly cystic Ovary Disorder (PCOD) is a hormonal imbalance in women that can cause irregular periods difficulties with fertility metabolic issues and long-term health risks. Self-Supervised Learning (SSL) is a type of machine learning where the model teaches itself from unlabeled data learning useful patterns and representations without needing manual labels. Deep Clustering is a method that combines deep learning and clustering allowing the model to group similar data points more effectively in a high dimensional feature space. SimCLR is a self-supervised learning framework that uses contrastive learning to make different augmented versions of the same data point similar while keeping different data points distinct. MoCo (Momentum Contrast) is an SSL technique that keeps a dynamic memory of representations to improve contrastive learning especially useful when batch sizes are small. Deep Cluster is an unsupervised method that alternates between clustering learned features and updating the neural network to improve the

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quality of the feature representations. SWAV is a clustering based contrastive method that learns feature representations while simultaneously assigning data points to clusters creating clear and well separated embedding.

II. LITERATURE SURVEY

Early studies used supervised ML methods such as Support Vector Machines and Random Forest to predict PCOD from clinical and hormonal datasets. While these approaches achieved high accuracy, they assumed binary outcomes and did not explore phenotype diversity. More recently ensemble methods and hybrid models have been applied showing improved detection but still lacking a focus on sub type discovery. Transfer learning approaches such as PCO Net leveraged pretrained convolution networks to detect PCOD in small datasets. Although effective in classification tasks they were not designed to uncover heterogeneity among patients. Self-supervised learning has emerged as a powerful paradigm in healthcare reducing the need for labeled data. SimCLR and MoCo adopt contrastive learning to create robust representations while DeepCluster and SwAV extend this by coupling representation learning with clustering These methods have been successfully applied in biomedical imaging cancer phenotype and electronic health records but remain under explored in PCOD. Recent reviews in reproductive medicine also highlight deep learning applications for gynecology and PCOS detection. Furthermore, clinical deployment of AI systems is gaining attention with decision support systems in reproductive endocrinology emerging as critical tools. However, adoption challenges remain particularly around explain ability and ethical safeguards. Our study fills this gap by applying SSL and clustering to clinical PCOD data enabling label free phenomapping and demonstrating deployment in a clinical decision support platform.

The reviewed studies collectively highlight the evolution of artificial intelligence (AI) and machine learning (ML) techniques applied to PCOD prediction and broader healthcare contexts. Early works like the 2018 *DeepCluster* and 2020 *SwAV* established foundational methods for self-supervised and contrastive clustering, forming the basis for later biomedical applications despite being trained on nonclinical datasets. Subsequent research in 2022 and 2023 advanced these concepts toward healthcare, with large-scale self-supervised learning (SSL) studies demonstrating scalability on multimodal datasets exceeding 100 million records—though such approaches demand heavy computational resources. Domain-specific studies, including PCOD prediction using SVM and Random Forest (2023) and ensemble learning (2021), achieved high accuracy on clinical data but were limited to binary classification without addressing subtype differentiation. The introduction of *PCONet* (2022), an Inception-based transfer learning framework, improved detection on small datasets, proving the usefulness of pretrained features though not tailored for PCOD subtypes. Meanwhile, reviews like *PCOS Diagnosis* (2022) and *Deep Learning in Gynecology* (2023) provided comprehensive overviews of ML applications in reproductive medicine, highlighting progress yet underscoring the absence of SSL-focused work. Broader surveys, such as those on SSL in healthcare (2023) and biomedicine (2024), justified the need for reduced labeling efforts and guided method selection across domains, but lacked PCOD-specific deployment insights. Complementary studies on SSL for time-series (2022) and EHR data (2022) further demonstrated robust embeddings relevant to PCOD biometrics. Finally, research addressing trustworthy AI (2023) and decision support systems in reproductive endocrinology (2023) emphasized transparency, fairness, and workflow integration—critical for deploying AI-based PCOD diagnostic systems—though often within limited case scopes or general healthcare contexts.

III. DATASET

The dataset used in this study consisted of anonymized patient records, each containing a combination of demographic, clinical, and biochemical information. Specifically, the features included Age, Body Mass Index (BMI), and key hormonal markers such as Luteinizing Hormone (LH), Follicle-Stimulating Hormone (FSH), Anti-Müllerian Hormone (AMH), and Insulin levels. Together, these parameters provide a comprehensive snapshot of both reproductive and metabolic health, making them highly relevant for stratifying patients into clinically meaningful PCOD subtypes. All patient data was collected directly through the Django-based web interface developed for this research. The platform allowed clinicians or researchers to securely enter patient information. Once the data was submitted, the backend system automatically processed the input, applied the trained self-supervised learning and clustering models, and generated a corresponding cluster assignment for each patient. This real-time integration ensured that the dataset was both clinically useful and continuously updated as new patients were added

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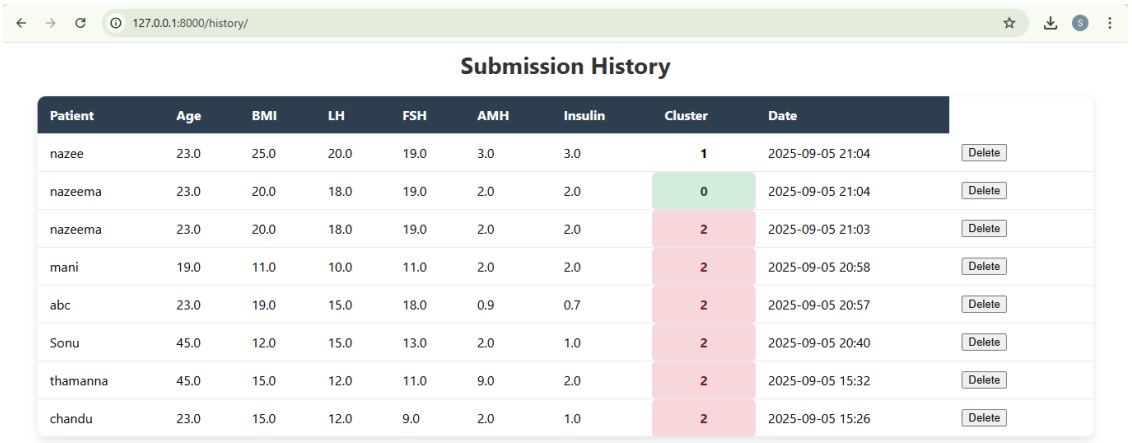

Fig. 1. Submission history with patient records and cluster assignments.

IV. METHODOLOGY

The proposed framework integrates SSL with deep clustering to uncover PCOD sub types. The overall pipeline is illustrated in Fig. 2.

Step 1: Data Pre-processing

Clinical features (age BMI LH FSH AMH insulin glucose lipid profile) are preprocessed using:
Missing value imputation with K nearest neighbors (KNN). Winsorization for outlier control. Min–max normalization to ensure comparable scales.

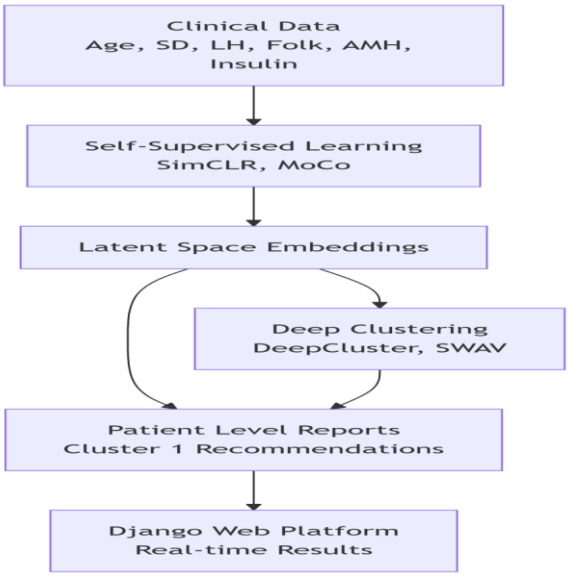

Fig. 2. End to end workflow of the proposed framework.

Step 2: Representation Learning with SSL

We adopt Sim CLR and MoCo to learn patient embed dings. Each record undergoes augmentation to create positive pairs while other patients act as negatives. The encoder network is optimized using the Info NCE loss:

L=−log Σk=1Nexp(sim(zizk)/τ)exp(sim(zizj)/τ)

Where:
zi,zjz_i, z_jzi,zj = embeddings of a positive pair (e.g., two different views of the same patient data)
zkz_kzk = embeddings of negative samples (e.g., data from other patients)
τ\tauτ = temperature parameter that controls how sharply the similarities are scaled
sim(a,b)\text{sim}(a,b)sim(a,b) = similarity function (often cosine similarity or dot product)

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The numerator: exp⁡(sim(zi,zj)/τ)\exp(\text{sim}(z_i, z_j)/\tau)exp(sim(zi,zj)/τ) Measures how similar the positive pair (same patient) is.
The denominator**:** Σk=1Nexp⁡(sim(zi,zk)/τ)\sum_{k=1}^{N} \exp(\text{sim}(z_i, z_k)/\tau)Σk=1Nexp(sim(zi,zk)/τ) Sums similarity of ziz_izi with all samples**,** including positives and negatives.

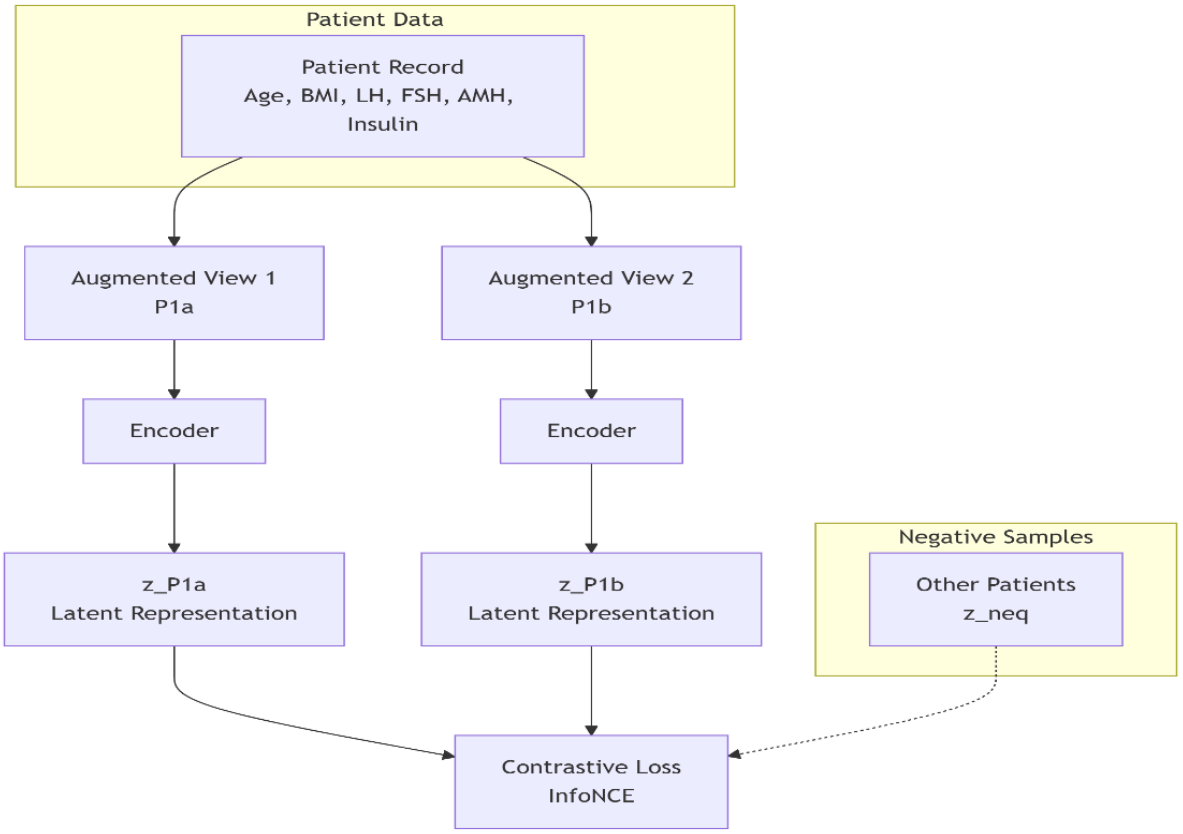

Fig. 3. Contrastive SSL for PCOD with Info NCE los

The log fraction ensures we want the positive pair similarity to dominate while negative samples get **s**maller weights**.**

Step 3: Deep Clustering
Latent embedding are clustered with Deep Cluster and SwAV. Deep Cluster alternates between k means assignment and encoder refinement while SwAV simultaneously learns prototypes and embedding. Cluster quality is evaluated using Silhouette Score and Davies Bouldering Index.
Step 4: Clinical Integration
The trained system is integrated into a Django platform enabling clinicians to:
Upload patient data.
View clustering results in real time.
Access visual analytic (pie/bar charts box plots).
Download patient level reports with phenotype meaning and recommendations.

V. RESULTS

A. Cluster Distribution

The framework identified three phenotypes: Cluster 0 (33.3%) Cluster 1 (44.4%) and Cluster 2 (22.2%) shown in Fig. 3.

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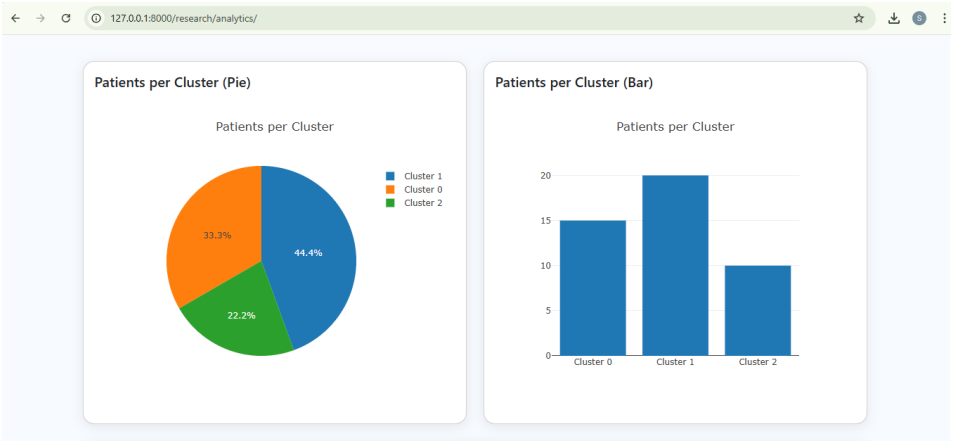

Fig. 4. Distribution of patients across clusters: (a) pie chart (b) bar chart.

B. Feature Differentiation

Box plots (Fig. 4) highlight clinical differences: Cluster 0 patients are near normal Cluster 1 shows hormonal irregularities and Cluster 2 displays insulin resistance.

Fig. 4. Box plots of features grouped by cluster.

C. Cluster Validity

Silhouette analysis confirmed K=3K=3K=3 as the optimal clustering (Fig. 5).

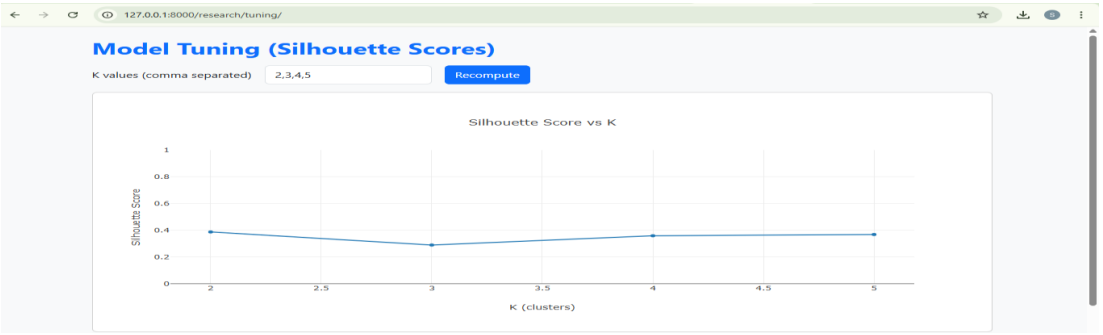

Fig. 5. Silhouette Score variation across different K values.

D. Patient Reports

Two sample patient reports are shown in Fig. 6:

(a) Sonu 45 years - severe phenotype requiring insulin management.

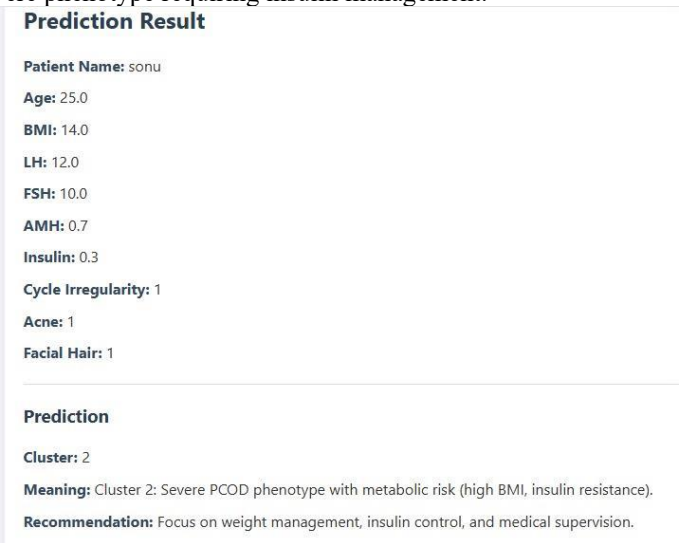

Fig. 6. Patient level reports with cluster prediction phenotype meaning and recommendations.

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(b) Nazeema 22 years -severe phenotype with hormonal irregularities.

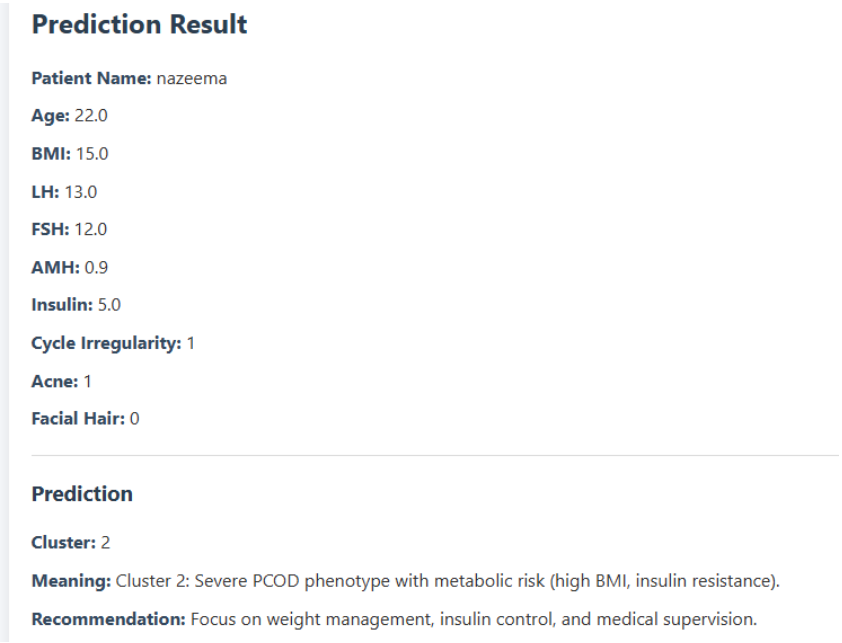

Fig. 7. Patient level reports with cluster prediction phenotype meaning and recommendations.

VI. DISCUSSION

The proposed framework demonstrates that self-supervised learning-based embedding combined with deep clustering can reveal clinically meaningful PCOD sub types. Unlike supervised methods this approach does not rely on labels making it more scalable for real world healthcare settings where annotation is often limited and expensive. Our study confirms that three distinct phenotype clusters emerge from the clinical data set aligning well with the spectrum of metabolic and hormonal irregularities described in the PCOD literature.

Importantly this work provides evidence that PCOD should not be reduced to a binary classification of presence or absence. Instead our findings suggest that patient specific variations in hormonal levels (e.g. LH/FSH ratio AMH concentration) and metabolic markers (e.g. insulin resistance lipid profile) can be leveraged to create more tailored treatment pathways. For example Cluster 0 patients may benefit from routine monitoring and lifestyle interventions while Cluster 2 patients may require early initiation of insulin sensitizing therapy. Such differentiation supports precision gynecology and aligns with current trends in individualized medicine.

When comparing our framework with existing machine learning studies in PCOD we observe a major methodological shift. Traditional SVM and Random Forest models achieved high predictive accuracy but were restricted to binary outcomes. Transfer learning approaches like PCO Net expanded this by leveraging pretrained models but still lacked sub type discovery. Our SSL driven method bridges this gap by both reducing labeling requirements and uncovering hidden sub types thereby offering clinical insights beyond prediction.

Moreover, the integration of our system into a Django based clinical decision support platform demonstrates the transnational potential of this work. By providing real time visualization of clusters clinicians can better understand patient stratification and generate reports that are actionable in practice. This user centered design is essential for building trust among healthcare professionals and accelerating adoption of AI driven tools in gynecology.

Beyond PCOD the methodological framework we present may generalize to other endocrine or metabolic disorders where heterogeneity is significant such as thyroid disorders metabolic syndrome and gestational diabetes. Future studies could explore the transfer ability of SSL embedding across such conditions.

VII. LIMITATIONS

The size of the dataset utilized in this study is rather small and only reflected a single-center collection of data, thereby limiting the generalizability of findings to broader populations with varied ethnicities, lifestyles, and genetics. While there were internal clustering validation metrics (e.g., Silhouette Score) that confirmed the stability of our findings, we

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still have limited evidence for reproducibility due to the absence of external validation across larger multi-center datasets. Moreover, while our clustering revealed different subtypes of our collected data, that does not provide graphical interpretability for SSL embeddings. Under circumstances of being in a high-stakes environment and the absence of explainability modules, we would expect clinicians have difficulties fully trusting or adopting these systems into practice. Additionally, since the data used herein potentially has bias, multiple imputation steps, along with preprocessing choices, may result in inaccuracies; for example, KNN imputation assumes patients are like each other which may not apply across heterogeneous populations. Another limitation is the restricted feature set; in this initial study we only included clinical and biochemical features and excluded genomic, imaging, and lifestyle-based data which may scrutinize phenotype diversity. Taken together, the computational cost of SSL frameworks like SimCLR and MoCo demands significant resources which while manageable for research may in turn be a barrier for smaller clinics implementing these models. Finally, ethical issues complicate implementation as AI-based patient Acknowledging these limitations provides direction for future work. Incorporating multi modal datasets enhancing explain ability features addressing ethical safeguards and validating across diverse populations will strengthen the clinical applicability of this approach.

VIII. CONCLUSION AND FUTURE WORK

This paper introduced a self-supervised and clustering based framework for PCOD phenomapping. Results confirmed three distinct phenotypes validated through clinical indicators. The deployment of a Django based platform demonstrated practical usability through real time predictions and patient level reports.

Contributions:
Contrastive SSL applied to PCOD clinical data.
Deep clustering-based phenotype discovery.
Clinician friendly deployment with real time reports.

Future Work:
Extend validation to multicenter datasets (>1000 patients).
Incorporate genomic and lifestyle data for richer phenotyping.
Add explain ability features to enhance clinician trust.
Explore multi modal integration with imaging and wearable.
Develop strategies for ethical deployment and clinician training to improve adoption.

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