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AI Models and Genomic Biomarkers in Rare Gynecological Pathologies: From Mayer-Rokitansky-Küster-Hauser (MRKH) and Primary Ovarian Insufficiency (POI) to Sex Cord-Stromal Tumors with Meigs' and OHVIRA Syndromes

Yash Srivastav[1*], Stuti Verma[2], Kamini Prajapati[1], Sandeep Prakash[1], Rajeev Kumar[2], Anubha Dhuriya[2], Anup Kumar Sirbaiya[3], Shivani Singh[1]

[1] D.K.R.R Pharmacy College (Dev Kumari Rajaram Pharmacy Shikshan Sansthan), Amberpur, Sitapur (Uttar Pradesh), India. 261303
[2] Aryakul College of Pharmacy and Research, Sitapur, Uttar Pradesh, India. 261303
[3]K.P. Singh Memorial Institute of Pharmacy, Sitapur, U.P, India.

***Corresponding Author E-mail: yashsrv.108@gmail.com**

Abstract

The rare gynecological diseases, including MRKH syndrome, POI, OHVIRA syndrome, Meigs' syndrome and SCSTs, pose great difficulties for diagnosis due to genetic complexity and clinical heterogeneity. This review highlights the latest human-based scientific evidence on the use of genomic biomarkers and AI in the diagnosis, prognosis, and treatment of the mentioned syndromes and disorders. The human studies performed through WGS, WES, next-generation sequencing, and multiomics revealed critical biomarkers associated with the development of reproductive tract disorders, ovarian function, and tumor progression, which promotes precision medicine and personalization. The review also covers the increasing usage of machine learning, deep learning, explainable AI, digital pathology, radiogenomics and AI-assisted diagnostics in analyzing genomic, imaging and histopathological data for accurate classification and diagnostics of diseases. Despite the high potential of these technologies in clinic, there are still some limitations related to the lack of large multicentric human datasets, genetic heterogeneity and the need for AI validation. Further studies should be directed at the combination of advanced genomic technologies, multiomics and explainable AI for the improvement of precision gynecology.

Keywords: Artificial Intelligence; Genomic Biomarkers; Rare Gynecological Disorders; Mayer–Rokitansky–Küster–Hauser (MRKH) Syndrome; Primary Ovarian Insufficiency (POI); Precision Gynecology

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1. INTRODUCTION

The rare gynecological pathologies are characterized by a wide range of conditions which affect the reproductive health of women, although having low incidence in nature. Rare gynecological pathologies include Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome, primary ovarian insufficiency (POI), obstructed hemivagina and ipsilateral renal anomaly (OHVIRA) syndrome, Meigs' syndrome, and Sex cord-stromal tumors (SCSTs). These diseases are generally related to such health problems as delayed diagnosis, infertility, menstrual disorders, endocrine dysfunction, pelvic pain and even malignant transformation[1]. Since these diseases share many symptoms, the conventional diagnostic approach cannot be effective enough due to the impossibility of early disease detection. There is a need to develop an innovative diagnostic approach that includes elements such as molecular biology, genomics, medical imaging, and computational intelligence.

Figure 1: Gynecological[2]

The recent progress of genomics medicine in the treatment of these rare diseases has been significant. The use of Whole Genome Sequencing (WGS), Whole Exome Sequencing (WES), targeted next-generation sequencing, chromosomal microarrays, transcriptomics, and multi-omics approaches in studies with humans has made possible the discovery of many different genetic mutations associated with the development of an embryo reproductive system, ovarian follicular development, DNA damage repair, and tumour development. Some of the genomics biomarkers, which include WNT4, HNF1B, LHX1, GREB1L, TBX6, BMP15, NOBOX, FIGLA, FSHR, MCM8, MCM9, STAG3, EIF4ENIF1, FOXL2, DICER1, TERT, and AKT1, have helped in expanding the knowledge of the pathogenesis of these rare diseases along with improved diagnosis and prognosis[3].

1.1 Background Information and Context

Availability of high-throughput sequencing technologies and computational methods has completely revolutionized the methods used in the diagnostics and treatment of rare gynecological diseases[4]. The traditional diagnostic process has relied heavily on clinical evaluation, radiology, histopathology, and hormone analysis; however, such techniques do not possess enough sensitivity to detect any underlying molecular pathologies. Studies in the field of human genomics have shown that most rare gynecological diseases exhibit unique genetic patterns that could be detected even before the onset of clinical symptoms. In addition, artificial intelligence-based methods have improved the analysis of complex clinical data through integration of genomics, radiology, pathology, and laboratory findings in comprehensive prediction models.

1.2 Objectives of the Review

The key goals of this literature review include:

- To present an overview of the existing evidence from human-based studies on the genomic biomarkers involved in the development of MRKH syndrome, Primary Ovarian Insufficiency, OHVIRA syndrome, Meigs' syndrome, and Sex Cord–Stromal Tumours.
- To study the impact of Whole Genome Sequencing, Whole Exome Sequencing, multiomics methods, and molecular profiling on the improvement of disease diagnosis and knowledge of its pathogenesis.
- To analyze the use of artificial intelligence (machine learning, deep learning, and explainable AI) in genomic analysis, medical imaging, digital pathology, and clinical decision-making.
- To highlight the latest trends of liquid biopsy, radiogenomics, digital pathology, AI-supported diagnosis, and precision medicine in rare gynecology.
- To determine the existing problems, research gaps, and future directions of genomics and AI in routine clinical practice in gynecology.

1.3 Importance of the Topic

Rare gynecological diseases constitute a major but often neglected issue in women's health care owing to their low frequency, complicated genetics, and delayed diagnosis. With the advances in genomics and artificial intelligence, the possibilities to address all those issues have been provided, allowing early diagnosis, accurate classification of the disease, personalized risk prediction, and tailor-made treatment[5]. The use of genomic markers together with AI algorithms holds a promise for improving reproductive prognosis, developing more efficient strategies for fertility protection, facilitating tumor diagnosis, and making decisions based on evidence. With the growing importance of precision medicine, the understanding of how genomics and artificial intelligence can work together in the field of rare gynecological diseases is essential.

2. GENOMIC BIOMARKERS IN RARE GYNECOLOGICAL PATHOLOGIES

Rare gynecological disorders are marked by large genetic and phenotypic diversity, which makes their diagnosis and treatment very complex. Recent developments in human genomics techniques such as whole exome sequencing (WES), whole genome sequencing (WGS), targeted NGS, RNA sequencing, and chromosomal microarray have significantly enhanced the identification of genetic mutations causing diseases[6]. These methods have facilitated the discovery of molecular biomarkers involved in the development of congenital reproductive system malformations, ovarian dysfunctions, and rare ovarian cancers. Human genomics research has helped achieve personalized medicine through early diagnosis, genetic counselling and tailored treatment of these diseases. The subsequent sections provide an overview of the most important genomic biomarkers involved in selected rare gynecological disorders, and their respective human studies, methodology used, main results, and limitations[7].

2.1 Genomic Biomarkers in Mayer–Rokitansky–Küster–Hauser (MRKH) Syndrome

The Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome represents a rare congenital condition that manifests itself as the absence or underdevelopment of the uterus and upper vagina in normally appearing females with a normal 46, XX chromosomal constitution[8]. Over the last 10 years, research in human genomics has greatly advanced knowledge of this condition. Using whole-exome sequencing and targeted next-generation sequencing approaches, several candidate genes such as WNT4, HNF1B, LHX1, GREB1L, and TBX6, involved in the development of the Müllerian duct and embryogenesis, have been implicated. Mutations in the WNT4 gene are known to cause cases of MRKH with an unusual phenotype of hyperandrogenism, while mutations in the LHX1 and HNF1B genes have been reported to cause reproductive abnormalities along with renal disorders.

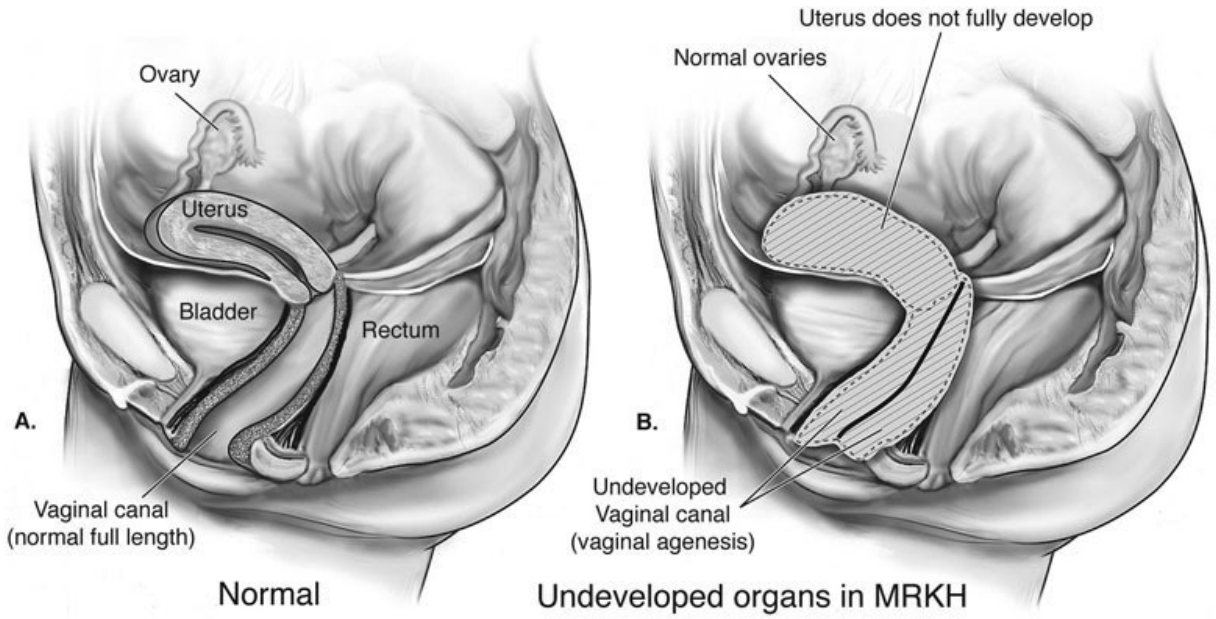

Figure 2: Mayer–Rokitansky–Küster–Hauser (MRKH)[9]

Human studies using copy number variation studies and chromosomal microarray technology have further shown that the condition known as MRKH syndrome is quite genetically heterogeneous, with no particular gene responsible for the bulk of the disorder. This implies that oligogenic inheritance, epigenetics, and environment factors could all play an important role in the disease development. Therefore, a combination of genomics approaches that include sequencing and transcriptomics is strongly advised[10].

2.2 Genomic Biomarkers in Primary Ovarian Insufficiency (POI)

Premature ovarian failure (POF), also known as primary ovarian insufficiency (POI), entails premature cessation of ovarian function prior to reaching 40 years and it has been described as one of the major genetic conditions causing female infertility[11]. Genomic studies conducted on humans have revealed a number of genes responsible for folliculogenesis, meiosis control, and DNA repair. The genes that have been reported as most frequent in their association with POI include BMP15, NOBOX, FIGLA, FSHR, MCM8, MCM9, STAG3, and EIF4ENIF1. BMP15, NOBOX, and FIGLA mutations affect oocyte maturation and folliculogenesis, while MCM8, MCM9, and STAG3 mutations affect DNA repair and chromosomal stability during meiosis.

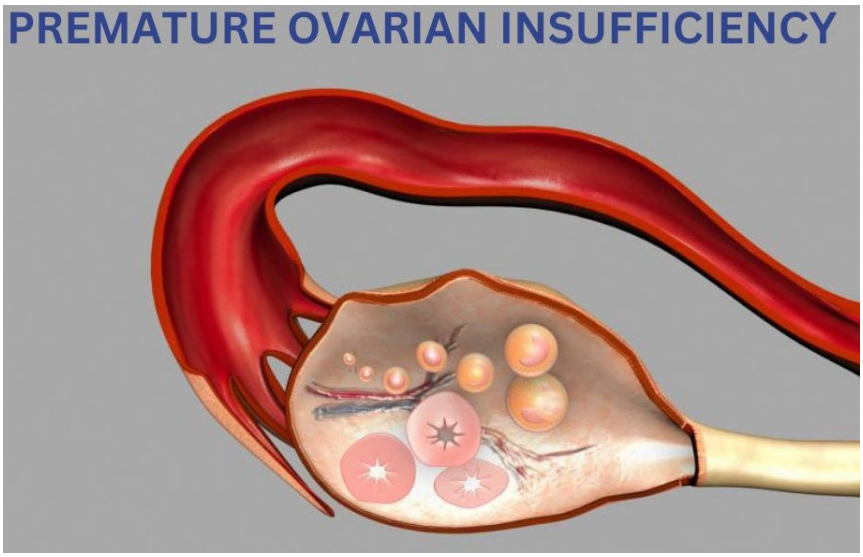

Figure 3: Primary ovarian insufficiency (POI)[12]

The diagnosis of women suffering from idiopathic POI has been markedly improved by large human cohorts through whole-exome sequencing and the role played by DNA repair pathways in POI has been proven to be a key factor in causing premature ovarian ageing[13]. However, many patients have remained undiagnosed genetically, which suggests that there are other genes and environmental factors contributing to the development of this condition.

2.3 Genomic Biomarkers in OHVIRA Syndrome

OHVIRA (Obstructed Hemivagina and Ipsilateral Renal Anomaly) syndrome refers to a congenital disorder characterized by the presence of uterine duplication, obstructed

hemivagina, and ipsilateral renal anomalies[14]. In humans, it has been repeatedly proven in clinical studies that MRI is the most effective way of diagnosis of the OHVIRA syndrome due to the high efficacy of visualization of the pelvic and urinary system anomalies. MRI findings have greatly improved preoperative planning and reduced diagnostic delays.

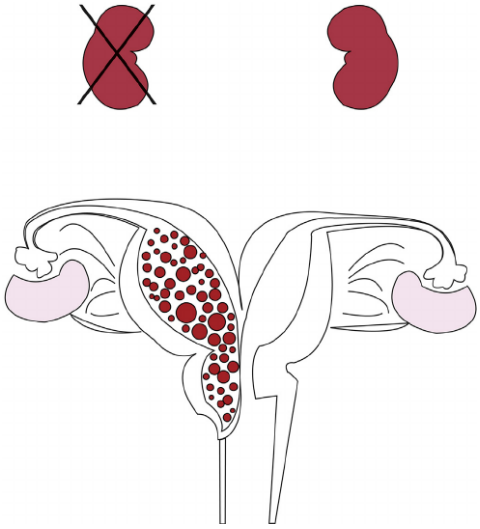

Figure 4: OHVIRA Syndrome[15]

As compared to MRKH syndrome, OHVIRA is less studied genotypically. Several studies involving humans have implicated a number of genes responsible for the development of the Müllerian and Wolffian ducts as factors that may cause susceptibility to OHVIRA, but a definite biomarker has not been found so far[16]. The evidence gathered by most researchers involves cases or small retrospective studies conducted in clinics.

2.4 Genomic Biomarkers in Sex Cord–Stromal Tumours

Sex-cord stromal tumors are rare forms of ovarian tumors which include granulosa cell tumors, Sertoli-Leydig cell tumors, fibromas, and thecomas[17]. Molecular pathological research has reported a number of genomic biomarkers that have diagnostic and prognostic importance. The FOXL2 mutation has been reported to be the molecular signature of granulosa cell tumors in adults, and its diagnostic sensitivity has also been established in human tumor specimens. Other molecular alterations such as DICER1, TERT, and AKT1 mutations have been linked to tumor progression and recurrence.

Recent studies on humans where targeted next generation sequencing is combined with histopathological and immunohistochemical studies have greatly enhanced the classification of tumours and prognostication. Molecular studies have also been instrumental in distinguishing between benign and malignant stromal tumours[18].

2.5 Clinical and Genomic Evidence in Meigs' Syndrome

Meigs' syndrome is a very rare benign syndrome that manifests as ovarian fibroma, ascites, and pleural effusion. Different from other rare gynecological syndromes, there has never been any

disease-specific biomarker discovered in human studies to this day[19]. The diagnosis is mostly based on clinical examination, imaging methods, such as ultrasound and MRI, tumour markers in blood, and finally histopathological examination after surgical removal of the tumour from the ovary. In human studies, complete surgical removal of the tumour typically causes regression of ascites and pleural effusion.

Despite the lack of molecular evidence, the progress in imaging and pathology has greatly helped in differential diagnosis of the disease and differentiating Meigs' syndrome from malignant ovarian tumors. Future multicentre research combining molecular analysis may lead to the discovery of new biomarkers[20].

3. AI MODELS IN RARE GYNECOLOGY

AI is one of the promising technologies in gynecology, which allows diagnosing, classifying, prognosticating and managing rare gynecological diseases. Implementation of ML, DL, and XAI algorithms combined with genomics sequencing, medical imaging, and digital pathology makes it possible to make more effective clinical decisions[21]. There have been numerous human studies that proved that AI algorithms were able to detect genomic patterns, predict disease outcomes, help in radiology interpretation and implement individualized treatments. Below there is a review of the current AI approaches that are studied for application in rare gynecological pathologies.

3.1 Machine Learning in Rare Gynecological Disorders

The machine learning algorithms have proven to be useful in analyzing data from clinical and genomics of rare gynecological diseases[22]. The studies conducted on humans have shown that supervised learning algorithms such as Random Forest (RF), Support Vector Machine (SVM), XGBoost, and Logistic Regression are capable of classifying the disease subtype, identifying genomic biomarker, and predicting clinical outcome.

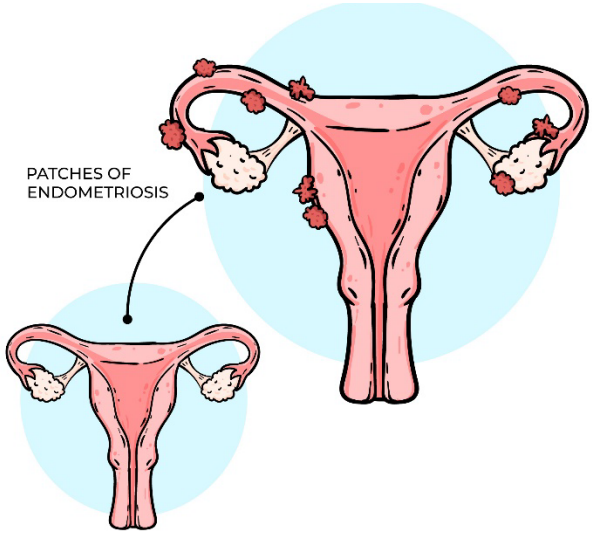

Figure 5: Gynecological Disorders[23]

Random Forest has found wide application in genomic data sets, owing to its capability of analysis of many genetic variables along with prevention from overfitting. The study conducted among humans with patients suffering from primary ovarian insufficiency and ovarian tumors revealed that Random Forest models are effective at recognizing candidate biomarkers in relation to ovarian dysfunction and tumor progression[24]. Support Vector Machine algorithms have shown high classification accuracy in differentiating between benign and malignant ovarian lesions by means of radiology and molecular markers. Likewise, XGBoost has been used for integrating genomic, laboratory, and clinical factors for disease prediction. Logistic Regression is still very important among statistical machine learning methods.

3.2 Deep Learning for Medical Imaging and Digital Pathology

Deep learning techniques have been quite useful in improving the accuracy of imaging diagnosis in cases of rare gynecological conditions through the processing of radiology and histopathology data sets[25]. Research involving humans has indicated that convolutional neural networks (CNNs) can diagnose such conditions effectively by analyzing the MRI and ultrasound images of congenital Müllerian anomalies, ovarian masses, and reproductive tract abnormalities. CNN-based models reduce observer variability while improving lesion detection and anatomical characterization.

Deep learning models have been effectively used in the field of digital pathology in order to automatically classify and grade tumours using whole-slide histopathological images. There have been human studies with regard to sex cord-stromal tumours which show that the CNN-based model has effectively helped in recognizing histopathological patterns and aiding pathologists in classifying the type of tumor[26]. In recent times, the use of Vision Transformers (ViT) has become common as an advanced deep learning architecture that can learn long-range features in images, and it has shown promising results in the domain of digital pathology.

3.3 Explainable Artificial Intelligence in Clinical Decision Support

Even though the predictive power of AI models is very high, the lack of transparency has prevented their adoption in clinical settings. Explainable Artificial Intelligence (XAI) solves this problem by offering clear explanations for AI predictions.

In human studies, it has been observed that explainable AI is able to improve the clinical decision support system by determining the factors that play a crucial role in the prediction of the diseases, including genomic, radiographic, and clinical factors. Explainable AI has also found application in predictive models in rare gynecologic diseases to predict fertility, ovarian cancer recurrence, and personalized medicine planning. There are also risk-stratification models that have used the genomic factors combined with clinical factors.

Through enhanced transparency and accountability, explainable AI helps foster cooperation between clinicians and artificial intelligence systems by enabling individualized treatment approaches. Nevertheless, additional validation is needed from multiple human data centers before these methodologies can be adopted in clinical settings.

4. RECENT DEVELOPMENTS

Genomic technology advancements in recent years have greatly improved the diagnostics and treatment of rare gynecological diseases due to the possibility of characterizing completely the genetics of disease-causing variants[27]. In particular, Whole Genome Sequencing (WGS) and Whole Exome Sequencing (WES) have become necessary tools in the identification of genetic pathogenic variations in such diseases as MRKH syndrome, POI, and rare sex cord-stromal tumors. Studies of human sequencing have discovered new variants within the genes responsible for the development of the Müllerian duct, folliculogenesis, and DNA repair. Moreover, multi-omics research methods have been introduced and helped achieve a better understanding of the molecular mechanisms behind these diseases by incorporating genomics, transcriptomics, epigenomics, proteomics, and metabolomics data analysis.

Other recent advancements have also illustrated the increasing relevance of liquid biopsy, radiogenomics, and digital pathology in precision gynecology. The use of liquid biopsy, which includes the evaluation of circulating tumour DNA (ctDNA), circulating tumour cells (CTCs), and extracellular vesicles, as a minimally invasive technique has been successful in the monitoring of tumour progression, recurrence, and assessment of treatment response in ovarian tumours. Likewise, radiogenomics has come to light as an innovative tool through the integration of magnetic resonance imaging (MRI) and other radiologic findings with genomics data[28]. Digital pathology has made it possible to use whole-slide imaging and automated image analysis to classify rare ovarian tumours more precisely based on histopathological criteria. Human studies have revealed that the use of imaging, pathology, and genomics increases the accuracy of diagnosis while decreasing inter-observer variability.

The incorporation of AI in genomics and clinical data is also contributing to the development of precision medicine in rare gynecology. Machine learning and deep learning models are gaining increasing popularity in analysing complex genomic datasets, predicting disease risks, classifying tumour types, and planning personalised therapies. AI-based diagnostic tools have proved more effective in analysing MRI, ultrasound, histopathological images and sequencing data, thus facilitating early diagnosis and accurate decision making. However, there are still numerous obstacles to overcome, such as limited access to large multicentric human datasets, variations in genomic results obtained among various populations, and the absence of unified validation standards[29]. The future research efforts should be directed at developing the use of advanced sequencing methods, multiomics analysis, explainable AI and international collaboration in order to set up precision medicine strategies in rare gynecological diseases.

Table 1: Summary of Selected Literature on AI Models and Genomic Biomarkers in Rare Gynecological Pathologies[30]

Author(s) & Year	Study Focus	Methodology/Model	Key Findings

Drymiotou et al. (2025)[31]	Molecular biomarkers in borderline ovarian tumors for personalized treatment and prognostic assessment	Literature review of human molecular, genomic, and clinical studies	Molecular biomarkers, including **KRAS, BRAF,** and **ERBB2** mutations, along with immunohistochemical markers, improved diagnostic accuracy, prognostic assessment, and personalized treatment strategies. However, larger multicentre clinical studies were recommended for biomarker validation.
Jain et al. (2025)[32]	Molecular diagnostics and pathology of gynecologic malignancies	Review of human pathological, genomic, and molecular diagnostic studies	Molecular biomarkers, next-generation sequencing, immunohistochemistry, and genomic profiling enhanced tumour diagnosis, classification, and prognostic evaluation, supporting precision medicine despite challenges related to standardization and accessibility.
Soleimani et al. (2025)[33]	Recent advances in early detection of ovarian cancer	Review of human clinical studies evaluating molecular biomarkers and liquid biopsy technologies	Circulating tumour DNA (ctDNA), circulating tumour cells (CTCs), extracellular vesicles, microRNAs, and protein biomarkers improved the sensitivity and specificity of early ovarian cancer detection, although further prospective multicentre validation was required.
Lee (2025)[34]	AI-based biomarker discovery for infertility-associated conditions (PCOS and recurrent implantation failure)	Human microbiome analysis integrated with machine learning algorithms	Artificial intelligence-assisted microbiome analysis improved biomarker identification, disease classification, and predictive modelling for infertility-related disorders, supporting personalized reproductive medicine.

Srivastava et al. (2026)[35]	Molecular pathology and diagnostics of gynecologic malignancies	Review of recent human clinical, molecular pathology, genomic, and AI-based diagnostic studies	Integration of genomic sequencing, molecular biomarkers, digital pathology, and AI enhanced tumour diagnosis, prognostic evaluation, and personalized treatment planning, while emphasizing the need for standardized validation and multicentre clinical implementation.

5. DISCUSSION

The section provides a critical evaluation of the evidence presented in this review in terms of the clinical relevance of the role of genomics and artificial intelligence in rare gynecological diseases. It also discusses how they affect precision medicine and highlights the current challenges and future research directions[36].

5.1 Interpretation and Analysis of Findings

According to the findings of this literature review, the introduction of genomic biomarkers along with the application of artificial intelligence (AI) technology has greatly facilitated the detection, diagnosis, classification, and treatment of rare gynecological pathologies. It is important to note that all human-based studies provide information that genomics methods such as Whole Exome Sequencing (WES), Whole Genome Sequencing (WGS), targeted next-generation sequencing, as well as chromosomal microarray, allowed to detect clinically significant biomarkers in case of such pathologies as Mayer–Rokitansky–Küster–Hauser (MRKH) syndrome, Primary Ovarian Insufficiency (POI), Sex Cord–Stromal Tumours, OHVIRA syndrome, and Meigs' syndrome[37]. Moreover, according to the reviewed literature, it is possible to conclude that the usage of artificial intelligence, including machine learning, deep learning, and explainable AI, allows interpreting better the data obtained during the process of genomics, radiology, and histopathology analyses. Thus, combining genomics with the AI-driven technology is a promising trend in precision gynecology and could facilitate clinical decisions making.

5.2 Clinical Significance and Current Challenges

The current studies have revealed a high clinical importance of the genomic markers and AI technology for the advancement in treating patients with rare diseases affecting the female reproductive system. Identification of the mutations in the genome allows for making an appropriate diagnosis, counseling, preserving fertility, and choosing an individual treatment approach. At the same time, the utilization of AI technology increases the accuracy of disease classification, tumor characterization, and prognosis[38]. Combining the genomic analysis with the application of more advanced imaging and pathology, as well as predictive analytics, has

helped to advance precision medicine and improve diagnostics. Nevertheless, several difficulties that prevent the wider application of the approach in clinics remain relevant, which include the rarity of the diseases, lack of big data from multiple centers, genetic heterogeneity of rare diseases, unclear clinical importance of many genomic mutations, and the inconsistency in the performance of AI models in various populations.

5.3 Research Gaps and Future Directions

Despite tremendous progress in the fields of genomic medicine and artificial intelligence, a number of significant research gaps are present. The majority of publications were carried out with the use of relatively small groups of patients, with retrospective and single-center designs, which limits the external validity of the results. Multimodal multiomics studies with genomics, transcriptomics, epigenomics, proteomics and metabolomics in patients with rare gynecological diseases are not widespread at all, and prospective multicenter validation of AI-assisted diagnostic tools is very scarce. In addition, the literature does not provide enough information about the long-term clinical efficacy, cost-effectiveness and implementation of precision medicine powered by artificial intelligence in the field of gynecology[39]. Therefore, future research should be oriented towards conducting of multicenter international studies with the involvement of such technologies as Whole Genome Sequencing, Whole Exome Sequencing, liquid biopsy, radiogenomics, digital pathology and explainable AI, and standardization of frameworks for genomic testing, AI validation, data governance and implementation of the technology into gynecologic practice. This will help to further advance precision gynecology[40].

6. CONCLUSION

This study is concluded with the opinion that the combination of genomic biomarkers and artificial intelligence (AI) has greatly contributed to the knowledge and treatment of rare gynecological diseases including Mayer–Rokitansky–Küster–Hauser (MRKH) syndrome, Primary Ovarian Insufficiency (POI), OHVIRA syndrome, Meigs' syndrome, and Sex Cord–Stromal Tumours. The human literature suggests that there has been great advancement in precision diagnosis by the use of genomic technologies such as Whole Genome Sequencing (WGS), Whole Exome Sequencing (WES), targeted sequencing, and multiomics, whereas AI techniques have greatly contributed in analysis of genomic, imaging and histopathological data for clinical decision making. However, limitations such as scarcity of multicentre human datasets, genetic heterogeneity, lack of standardization of validation methods, and ethical issues still hinder the wide implementation of these technologies. Therefore, future research must concentrate on large-scale human collaborative studies, AI with genomics technologies and standardized precision medicine approach to improve the early diagnosis, personalized treatment strategies and overall reproductive and clinical outcomes.

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