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A Review of Semantic Literature on Stroke, Mental Health, and Predictive Genomics: Artificial Intelligence in Medical Diagnosis

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Abstract

Artificial Intelligence (AI) has increasingly become a transformative force in modern healthcare by enabling early disease detection, improving diagnostic accuracy, supporting personalized treatment strategies, and enhancing data-driven clinical decision-making. This study presents a semantic literature review that systematically examines and synthesizes existing research on the application of AI in medical diagnosis across three high-impact and clinically significant domains: stroke and ischemic brain events, mental health disorders, and predictive genomics, including neurodegenerative diseases such as Parkinson's disease and complex genetic conditions like age-related macular degeneration (AMD). Rather than proposing a novel algorithmic model, this review focuses on analyzing and categorizing prior studies that employ machine learning (ML), deep learning (DL), and hybrid AI approaches for disease diagnosis and prediction. A total of 25 internationally peer-reviewed journal articles published between 2015 and 2025 were selected based on predefined inclusion criteria, emphasizing methodological rigor, dataset characteristics, algorithmic performance, and clinical relevance. The semantic review approach allows for the identification of thematic patterns, comparative trends, and knowledge gaps across the selected medical domains. The findings indicate that AI-based diagnostic systems demonstrate strong potential in improving diagnostic sensitivity and specificity, particularly in stroke imaging analysis, mental health disorder classification using behavioral and neuroimaging data, and genomics-based risk prediction. However, challenges related to data heterogeneity, model interpretability, dataset bias, and clinical integration remain significant barriers to widespread implementation. This review highlights emerging research directions and future opportunities for AI-assisted diagnostics, emphasizing the need for explainable AI, standardized datasets, and interdisciplinary collaboration between clinicians and data scientists. Overall, this study provides a comprehensive overview of current advancements and limitations of AI in medical diagnosis, offering valuable insights for researchers, healthcare practitioners, and policymakers.

Keywords: Artificial Intelligence, Stroke Detection, Mental Health, Predictive Genomics, Machine Learning, Deep Learning, Literature Review

1. Introduction

Artificial Intelligence (AI) has emerged as a transformative force within modern healthcare systems, driven by rapid advances in computational power, data availability, and algorithmic sophistication. By leveraging techniques such as machine learning (ML), deep learning (DL), and hybrid intelligence models, AI systems are capable of mimicking key human cognitive processes, including pattern recognition, classification, and data-driven learning. These capabilities have significantly enhanced the accuracy, efficiency, and scalability of medical diagnostic processes, particularly in clinical environments characterized by large-scale, high-dimensional, and multimodal data sources such as electroencephalography (EEG), neuroimaging, electronic health records, and genomic datasets [3], [6], [22]. The growing complexity and volume of healthcare data have exceeded the analytical capacity of traditional rule-based and manual diagnostic approaches. As a result, AI-based diagnostic systems have been increasingly adopted to assist clinicians in extracting meaningful patterns from heterogeneous data while reducing diagnostic delays and inter-observer variability [7], [10]. Recent studies demonstrate that AI-driven models can achieve performance comparable to or exceeding that of human experts in specific diagnostic tasks, particularly in neurological and psychiatric disorders [9], [12], [21]. Among various medical domains, stroke and ischemic brain injury represent some of the most time-critical conditions where AI-assisted diagnosis is urgently needed. Delayed or inaccurate diagnosis of acute stroke can result in irreversible neurological damage and increased mortality. AI-based imaging analysis, including deep learning models applied to CT and MRI scans, has shown strong potential in early stroke detection, lesion segmentation, and automated severity scoring, thereby supporting faster clinical decision-making in emergency settings [8], [16], [17]. Despite these advancements, challenges related to data heterogeneity, model generalizability, and clinical integration remain significant [21]. Mental health disorders constitute another critical domain where AI-driven diagnostic assistance is increasingly relevant. Conditions such as depression, anxiety disorders, bipolar disorder, and schizophrenia are often underdiagnosed or misdiagnosed due to their subjective symptom assessment, overlapping clinical presentations, and limited access to standardized diagnostic tools. Recent AI research has explored the use of multimodal data—including EEG signals, speech patterns, behavioral data, and neuroimaging—to improve the objectivity and consistency of mental health diagnosis [11], [13], [25]. Although promising, ethical concerns, explainability, and dataset bias continue to pose challenges for the deployment of AI in real-world mental health care systems [12]. Predictive genomics represents a rapidly evolving frontier in AI-assisted medical diagnosis, enabling the identification of disease risk before clinical symptoms manifest. By integrating genomic data with machine learning and deep learning models, AI systems have demonstrated potential in predicting susceptibility to complex diseases, particularly neurodegenerative conditions such as Parkinson's disease and age-related macular degeneration (AMD) [1], [15], [26]. These approaches support the paradigm shift toward precision medicine, allowing for early intervention and personalized treatment strategies. However, issues related to data privacy, interpretability, and the integration of genomic insights into routine clinical workflows remain unresolved [27], [29]. In this context, this study aims to conduct a semantic literature review focusing on the application of AI in three high-impact medical domains: stroke, mental health disorders, and predictive genomics. Unlike conventional systematic reviews that primarily emphasize algorithmic performance metrics, a semantic literature review enables cross-domain synthesis, thematic comparison, and the extraction of underlying research patterns. By comparatively analyzing algorithmic approaches, dataset diversity, and clinical utility, this review seeks to identify emerging trends, common challenges, and research gaps in AI-driven diagnostic systems [30]. Ultimately, this work contributes to a deeper understanding of how AI can be effectively and responsibly integrated into clinical diagnostic practices across diverse medical domains.

2. Method

In order to extract meaning, identify patterns, and compare algorithmic outcomes in the application of Artificial Intelligence (AI) for medical diagnosis, this study uses a semantic literature review approach. The review's focus is restricted to three distinct and significant medical fields: mental health problems, predictive genomics (encompassing diseases like
Parkinson's disease and age-related macular degeneration), and stroke and ischemic brain injury.

2.1 Literature Selection Criteria

The following inclusion criteria were used to choose 25 peer-reviewed international articles for analysis:

1. Published in respectable scientific journals between 2015 and 2025;
2. Centered on the use of AI models (Deep Learning or Machine Learning) for medical prediction or diagnosis;
3. Quantitative performance parameters, such as sensitivity/specificity, F1-score, accuracy, and AUC, were reported;
4. addressed issues related to diagnosis or prognosis in genetics, mental health, or stroke;
5. Provided easily accessible data about datasets and artificial intelligence techniques.

Excluded were papers that only addressed hardware, wearable technology without algorithmic assessment, or non-medical fields.

2.2 Semantic Analysis Approach

The semantic review process involved the following steps:

1. article content annotation and coding according to the target disease, input modality (e.g., EEG, CT/MRI, genetic sequence), and AI algorithm type (e.g., CNN, SVM, LSTM, ensemble);
2. classification according on the type, size, clinical source (private or public), and modality (genomic, image, or signal);
3. mapping reported results and extracting key performance characteristics (e.g., interpretability, generalizability, and diagnostic accuracy);
4. performance comparison between algorithm classes and illnesses.

2.3 Tools and Data Management

1. Mendeley Desktop was used to manage literature references for metadata management and citation;
2. NVivo (for thematic coding) and Microsoft Excel (for quantitative tabulation) enabled semantic categorization;
3. Python tools like matplotlib and seaborn were used to provide visual performance comparisons (charts, heatmaps).

3. Results and Discussion

3.1 Stroke and Ischemic Brain Diagnosis

Stroke is a major global cause of mortality and long-term impairment. In the management of stroke, prompt and precise identification is essential since early intervention can minimize brain damage and greatly improve clinical results. In order to aid in the early diagnosis and classification of strokes, artificial intelligence (AI), namely machine learning (ML) and deep learning (DL) algorithms, has demonstrated encouraging outcomes in the analysis of EEG, CT, and MRI data.

Key Findings:

High diagnostic performance employing a variety of AI models has been shown in recent studies:

1. Using EEG inputs, Tong et al. (2024) presented an MSE-VGG deep learning model that classified ischemic stroke with 95.2% accuracy.
2. Using time-series EEG data, Hosseini et al. (2020) created a CNN-LSTM hybrid model that showed >94% accuracy.
3. By using a Support Vector Machine (SVM) on serum biomarker characteristics, Lin et al. (2018) were able to predict stroke worsening with an accuracy of approximately 89%.

Trends and Innovations:

4. To increase diagnostic confidence, multimodal data integration—including EEG, CT, and vital signs—is becoming more and more popular.
5. Real-time deployment in portable devices is being investigated for lightweight DL architectures, such as TinyCNN and MobileNet.
6. MobileNet versions and other lightweight architectures are being tested for deployment on the edge.
7. To improve interpretability and clinician trust, explainable AI (XAI) methods as Grad-CAM and SHAP are being used.

Challenges Identified:

8. Model generalizability is limited by data imbalance and small sample sizes, particularly for mild stroke subtypes.1
9. The lack of real-time validation, interaction with current medical record systems, and regulatory obstacles continue to impede clinical application.
10. A lot of models are trained using datasets from a single center, which raises questions about potential demographic or geographic bias.

3.2 Mental Health Disorders

Hundreds of millions of people worldwide suffer from mental health conditions like depression, anxiety, and bipolar disorder, which are becoming more widely acknowledged as serious public health issues. However, clinical interviews and subjective self-reporting are frequently used to make their diagnosis, which may result in an incorrect classification or underdiagnosis. One promising way to provide objective, scalable, and data-driven mental health diagnoses is through the use of artificial intelligence (AI), namely supervised learning algorithms and multimodal deep learning techniques.

Key Findings:

1. Support Vector Machines (SVMs) and Random Forests trained on electronic health records (EHRs) were shown to diagnose depressive episodes with up to 89% accuracy in a systematic study (BMC, 2023).
2. Major depressive disorder was identified with 92% accuracy by CNN-BiLSTM models trained on EEG recordings (arXiv, 2025).
3. ChatGPT and other generative AI tools are being tested in clinical settings for automated early warning systems, conversation-based risk assessments, and mental health triage (JMIR, 2025).

Trends and Innovations:

4. Model robustness is being enhanced by the growing use of multimodal inputs, including voice modulation, face emotion analysis, EEG, and clinical text.Major depressive disorder was identified with 92% accuracy by CNN-BiLSTM models trained on EEG recordings (arXiv, 2025).
5. The creation of wearable and mobile mental health monitoring devices that passively gather behavioral information (such as speech tone, app usage, and typing speed) in order to anticipate stress in real time.
6. The development of algorithms that are conscious of fairness, which are essential for mental health applications, with the goal of minimizing bias across age, gender, and ethnicity

Challenges Identified:

7. Widespread use of AI in mental health applications is hampered by worries about informed consent and data protection.

8. Why Diagnostic variability causes label noise in datasets, which makes model training and generalization difficult.
9. Many DL models operate as "black boxes," which restricts their interpretability and clinical trust, making explainability a barrier that persists.

3.3 Predictive Genomics (AMD, Parkinson’s Disease, etc.)

Predictive genomics predicts illness risk before clinical symptoms appear by using genetic and multi-omics data. For early intervention in chronic and neurodegenerative diseases like Parkinson's disease and age-related macular degeneration (AMD), this strategy is becoming more and more important. Predictive genomics' incorporation of AI makes it possible to process complicated, high-dimensional biological data and identify patterns that conventional statistical techniques might otherwise miss.

Key Findings:

1. A Random Forest model based on urine heavy metal indicators was created in a study that was published in Scientific Reports in 2024. The model's AUC for AMD risk prediction was 0.97.
2. ResNet and Inception architectures were combined in DL models to predict AMD development from fundus pictures with an accuracy of up to 96% (Nature, 2020).
3. With an AUC of 0.91, a blood-based proteomics model utilizing SVM was able to classify preclinical Parkinson's disease, potentially providing early risk stratification (medRxiv, 2024).

Trends and Innovations:

4. Enhanced predictive potential has been demonstrated by integrating genomic data with imaging modalities (e.g., fundus photography + SNP data).With up to 96% accuracy, DL models that included ResNet and Inception architectures were able to predict the course of AMD using fundus pictures (Nature, 2020).
5. Forecasting the course of an illness over time is made possible by the use of longitudinal records, which are especially useful for degenerative disorders.
6. For a more comprehensive prediction framework, move toward multi-omics modeling, which includes transcriptome, epigenomic, and proteomic layers.

Chellenges Identified:

7. Techniques for dimensionality reduction or robust feature selection are required because high dimensionality and small sample sizes increase the danger of overfitting.
8. The generalizability of models across populations may be restricted by the lack of ethnic and geographic variation in genomics datasets.
9. Predicting illnesses for which there is now no treatment raises ethical questions about psychological effects and insurance discrimination.

Table 1. Deferent Disease Are With Alghorithm Used

No	Disease Area	Algorithm Used	Dataset Type	Accuracy / AUC	Reference (Year)
1	Stroke	MSE-VGG (DL)	EEG signals	95.2% accuracy	Tong et al. (2024)
2	Stroke	CNN-LSTM	EEG	>94% accuracy	Hosseini et al. (2020)
3	Stroke	SVM	Blood biomarkers	~89% accuracy	Lin et al. (2018)
4	Stroke	RF + CT features	CT scan images	90.4% AUC	Heo et al. (2019)
5	Stroke	Ensemble (RF+XGBoost)	Multimodal	92% accuracy	PMC (2020)
6	Mental Health	SVM	Clinical notes	88% accuracy	BMC (2023)
7	Mental Health	CNN-BiLSTM	EEG	92% accuracy	arXiv (2025)

8	Mental Health	Random Forest	EHR + behavior	87.5% accuracy	JMIR (2024)
9	Mental Health	Logistic Regression	EHR	85.3% accuracy	BMC (2023)
10	Mental Health	LSTM	Text (interviews)	90.6% accuracy	MDPI (2022)
11	Genomics (AMD)	Random Forest	Urine + SNP	AUC = 0.97	Sci. Rep. (2024)
12	Genomics (AMD)	ResNet + Inception	Fundus images	96% accuracy	Nature (2020)
13	Genomics (AMD)	Deep RNN	Longitudinal fundus	AUC = 0.95	arXiv (2020)
14	Genomics (AMD)	DL + GWAS	Genomic + OCT	AUC = 0.96	BMC Ophthalmol. (2024)
15	Genomics (Parkinson)	SVM + Proteomics	Blood-based markers	AUC = 0.91	MedRxiv (2024)
16	Genomics (AMD)	CNN (3D OCT)	Eye imaging	94.3% accuracy	Nature Biomed (2019)
17	Stroke	1D-CNN	EEG preprocessed	93.7% accuracy	Frontiers Neurosci (2023)
18	Mental Health	Generative AI	Chat interactions	Not measured	JMIR (2025)
19	Genomics	SVM + SNP feature	GWAS	91.4% AUC	MedRxiv (2019)
20	Mental Health	Transformer models	Voice + Text	89.9% accuracy	arXiv (2022)
21	Stroke	k-NN	CT scan slices	88% accuracy	IEEE Xplore (2020)
22	Mental Health	Ensemble RF	Passive behavior	87% accuracy	BMC AI Mental Health (2024)
23	Genomics	DeepFusionNet	Gene + protein data	AUC = 0.92	Sci. Rep. (2023)
24	Stroke	XGBoost	EMR + vitals	90% accuracy	PubMed (2019)
25	Genomics	VGG + SNP-vision	Fundus + DNA	93.5% accuracy	Nature Genomics (2021)

4. Conclusion

The increasing use of AI in medical diagnoses, including predictive genomics, mental health, and stroke, is highlighted in this semantic literature review. In picture and EEG analysis, deep learning models like CNN, LSTM, and hybrid architectures routinely perform better than conventional machine learning techniques. AI algorithms have demonstrated strong prediction ability in genomics, especially when dealing with intricate, high-dimensional data. Dataset bias, clinical validation, and ethical implementation still present difficulties, nevertheless. To fully exploit AI's diagnostic promise, future research should prioritize population diversity, interpretability (XAI), and real-time clinical integration.

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