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ARTIFICIAL INTELLIGENCE IN EARLY DIAGNOSIS OF NEURODEGENERATIVE DISEASES: CURRENT EVIDENCE AND PROSPECTS

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Abstract
Neurodegenerative diseases, including Alzheimer's disease (AD), Parkinson's disease (PD), and amyotrophic lateral sclerosis (ALS), represent a growing global health burden, with millions of individuals affected worldwide and limited disease-modifying therapies available. Early and accurate diagnosis is critical for improving patient outcomes, yet conventional diagnostic approaches often fail to detect pathological changes prior to overt symptom onset. Artificial intelligence (AI), particularly machine learning (ML) and deep learning (DL) methodologies, has demonstrated remarkable promise in transforming early diagnostic paradigms through the analysis of neuroimaging, genetic, clinical, and digital biomarker data.

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This review synthesizes current evidence on AI applications in the early detection and differential diagnosis of major neurodegenerative conditions, examining the types of data modalities employed, algorithmic approaches, and reported diagnostic performance metrics. Convolutional neural networks (CNNs) applied to structural and functional MRI, PET imaging, and retinal scans have achieved diagnostic accuracies exceeding 90% in distinguishing AD from normal aging and mild cognitive impairment (MCI). Natural language processing (NLP) tools and speech analysis algorithms have been applied to detect subtle linguistic and acoustic markers predictive of neurodegeneration years before clinical diagnosis. Despite these advances, significant challenges persist, including limited dataset diversity, lack of prospective validation, interpretability concerns, and ethical issues surrounding data privacy and algorithmic bias. Federated learning and explainable AI frameworks offer potential solutions to enhance generalizability and clinical trustworthiness. The integration of multimodal AI platforms into clinical workflows requires interdisciplinary collaboration among neurologists, data scientists, ethicists, and policymakers. This review underscores the transformative potential of AI for early neurodegenerative disease diagnosis while emphasizing the necessity for rigorous validation, regulatory oversight, and equitable implementation to realize clinical benefits.

Introduction

Neurodegenerative diseases constitute a heterogeneous group of disorders characterized by the progressive loss of neuronal structure and function, leading to cognitive decline, motor dysfunction, and ultimately premature death [1, 4]. Globally, over 55 million individuals are estimated to live with dementia, the majority due to Alzheimer's disease, with projections indicating a tripling of this number by 2050 in the absence of effective preventive or therapeutic strategies [3]. Parkinson's disease affects approximately 10 million people worldwide, while ALS, though less prevalent, carries a particularly poor prognosis with median survival of two to five years from symptom onset [2, 7].

A fundamental challenge in neurodegenerative disease management is the substantial delay between pathophysiological onset and clinical recognition. Neuropathological hallmarks such as amyloid-beta plaques, tau neurofibrillary tangles, and alpha-synuclein aggregates may accumulate for decades before overt

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symptoms manifest [5, 12]. By the time a clinical diagnosis is established, significant and often irreversible neuronal loss has already occurred, severely limiting the window for therapeutic intervention [1, 8].

Artificial intelligence has emerged as a transformative tool in medical diagnostics, capable of identifying complex, multivariate patterns in large datasets that may be imperceptible to human clinicians [6, 14]. AI encompasses a range of methodologies, from classical machine learning algorithms such as support vector machines and random forests to sophisticated deep learning architectures including convolutional neural networks, recurrent neural networks, and transformer-based models [9, 14]. These approaches have been applied to diverse data types relevant to neurodegenerative disease diagnosis, including neuroimaging, genomics, proteomics, digital biomarkers, electronic health records, and wearable sensor data [6, 10, 16].

This review aims to provide a comprehensive synthesis of AI applications in the early diagnosis of neurodegenerative diseases, with emphasis on methodological approaches, clinical evidence, diagnostic performance, and translational challenges. By critically examining the current state of the art, we seek to identify priorities for future research and the path toward clinically viable, equitable, and interpretable AI-assisted diagnostics.

Neuropathological Basis and Biomarkers of Neurodegeneration

Understanding the molecular and cellular substrates of neurodegeneration is essential for contextualizing the biomarkers targeted by AI diagnostic systems. In Alzheimer's disease, the amyloid cascade hypothesis posits that aberrant processing of amyloid precursor protein leads to accumulation of amyloid-beta (Abeta) oligomers and plaques, triggering tau hyperphosphorylation, neurofibrillary tangle formation, synaptic dysfunction, neuroinflammation, and neuronal death [5, 12]. The preclinical phase, characterized by measurable amyloid and tau pathology in the absence of cognitive symptoms, may precede clinical onset by 15 to 20 years, providing a critical window for early intervention [3, 8].

Parkinson's disease is defined by the loss of dopaminergic neurons in the substantia nigra pars compacta and the presence of intraneuronal inclusions, termed Lewy bodies, composed principally of misfolded alpha-synuclein [2, 11]. Prodromal manifestations, including rapid eye movement sleep behavior disorder, anosmia, autonomic dysfunction, and subtle motor and cognitive changes, may antedate motor diagnosis by a decade or more, offering opportunities for presymptomatic detection [7, 11].

Established biomarkers for AI-based diagnostic models include cerebrospinal fluid (CSF) levels of Abeta42, total tau, and phosphorylated tau in AD; dopamine transporter imaging and skin biopsy alpha-synuclein in PD; and neurofilament light chain (NfL) across multiple neurodegenerative conditions as a marker of axonal injury [4, 8, 12]. Emerging blood-based biomarkers, including plasma Abeta42/40 ratios, phospho-tau217, and glial fibrillary acidic protein, have shown strong correlation with CSF and PET biomarkers, enabling minimally invasive large-scale screening amenable to AI integration [3, 5].

Artificial Intelligence Methodologies in Neurological Diagnosis

Machine learning and deep learning constitute the primary AI paradigms applied to neurodegenerative disease diagnosis. Supervised ML algorithms, including support vector machines, random forests, and gradient boosting, have been widely employed for classification tasks using structured clinical, neuropsychological, and biomarker data, achieving cross-validated accuracies of 80 to 92% in distinguishing AD, MCI, and healthy controls in large cohort datasets [9, 14, 16].

Deep learning, particularly convolutional neural networks, has revolutionized neuroimaging analysis by enabling automated, end-to-end feature extraction from raw image data without manual feature engineering [6, 13]. CNN-based models trained on structural MRI data have demonstrated sensitivity and specificity exceeding 90% for AD diagnosis, outperforming conventional volumetric analyses and radiologist assessments in several studies [13, 15]. Transfer learning, leveraging pre-trained networks such as VGG, ResNet, and Inception architectures on large general image datasets, has overcome limitations of relatively small neuroimaging cohorts, improving model generalization [6, 9].

Graph neural networks and attention-based transformer models have been applied to functional connectivity data derived from resting-state fMRI, capturing complex network topology alterations characteristic of neurodegeneration [10, 16]. Recurrent neural networks and long short-term memory architectures have been utilized for longitudinal data analysis, modeling disease trajectories and predicting progression from MCI to AD [14, 16]. Natural language processing algorithms analyzing transcribed speech samples, digital cognitive tests, and clinical notes have demonstrated capacity to detect linguistic markers of cognitive decline with diagnostic accuracy comparable to validated neuropsychological instruments [6, 15].

AI in Neuroimaging for Neurodegenerative Disease Detection

Neuroimaging represents the most extensively investigated data modality for AI-assisted neurodegenerative disease diagnosis. Structural MRI enables quantification of regional brain atrophy patterns, with AI models effectively identifying hippocampal, entorhinal cortical, and parietal volume reductions characteristic of AD with high reproducibility [13, 15]. Longitudinal MRI studies analyzed with AI have demonstrated capacity to predict conversion from amnestic MCI to AD with accuracy exceeding 85% up to three years before clinical diagnosis [8, 13].

Amyloid and tau PET imaging provide direct visualization of proteinopathies and represent gold-standard biomarkers for in vivo AD diagnosis. AI-based automated quantification of PET uptake patterns has shown concordance with expert visual reads while substantially reducing inter-rater variability and analysis time [5, 12]. Deep learning models combining amyloid PET with structural MRI have achieved area under the receiver operating characteristic curve values of 0.93 to 0.97 in predicting tau positivity and clinical progression [3, 12].

Dopamine transporter SPECT imaging analyzed with AI has improved diagnostic accuracy for Parkinson's disease and differentiation from essential tremor and other parkinsonian syndromes [7, 11]. Notably, AI analysis of retinal optical coherence tomography images has emerged as a promising non-invasive approach, exploiting the retina as a window to the brain, with models detecting neurodegenerative changes predictive of AD and PD from retinal structural and vascular alterations [4, 10].

Digital Biomarkers and Multimodal AI Approaches

Digital biomarkers derived from wearable sensors, smartphones, and remote monitoring platforms represent a rapidly evolving domain for passive, continuous, and ecologically valid assessment of neurodegenerative disease risk and progression [2, 16]. Gait analysis using inertial measurement units and computer vision has demonstrated discriminative power for PD, with AI models

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detecting characteristic bradykinesia, rigidity, and postural instability from accelerometer and gyroscope data with high sensitivity [7, 11]. Handwriting and drawing analysis through digitizing tablets, analyzed with ML, has shown diagnostic utility in PD and AD, capturing tremor, micrographia, and visuospatial deficits [2, 4].

Speech and language analysis represents a particularly promising non-invasive approach, given that vocal and linguistic changes may precede clinical diagnosis by years [6, 15]. AI models analyzing acoustic features including pitch variability, articulation rate, pause frequency, and vocal tremor have achieved diagnostic sensitivity of 80 to 95% for PD and AD from brief speech recordings [15]. NLP algorithms processing transcribed picture description tasks or spontaneous speech have identified semantic, syntactic, and pragmatic markers of cognitive decline, enabling remote and scalable cognitive screening [6].

Multimodal AI frameworks integrating neuroimaging, genetic, biomarker, clinical, and digital data have consistently outperformed unimodal approaches across diagnostic tasks [9, 14, 16]. Ensemble methods and data fusion architectures that combine predictions from specialized models trained on individual data streams have achieved diagnostic accuracies of 92 to 97% in differentiation of AD subtypes and identification of individuals at highest risk of progression [14, 16]. The integration of polygenic risk scores, derived from genome-wide association studies and processed with ML, with clinical and imaging data further enhances predictive power, particularly for sporadic disease [1, 9].

Clinical Evidence and Diagnostic Performance

Multiple large-scale studies utilizing established cohorts, including the Alzheimer's Disease Neuroimaging Initiative (ADNI), UK Biobank, and the Parkinson's Progression Markers Initiative (PPMI), have provided the primary evidence base for AI diagnostic models in neurodegeneration [3, 8, 11]. In the ADNI cohort, deep learning models combining MRI, PET, CSF, genetic, and cognitive data predicted AD diagnosis up to six years before clinical assessment with cross-validated accuracy of 84 to 92% [8, 13].

A systematic review of 50 studies applying ML to AD diagnosis reported a pooled sensitivity of 89% and specificity of 85% for distinguishing AD from healthy controls, with superior performance observed for deep learning relative to

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conventional ML approaches and for multimodal relative to unimodal data integration [14]. For PD, AI models applied to DaTSCAN imaging achieved diagnostic accuracy of 96%, while wearable-based models demonstrated 87% accuracy for prodromal PD detection in high-risk cohorts [7, 11].

Clinical deployment studies, though fewer, have demonstrated feasibility of AI integration into specialist memory clinic workflows. An AI decision support system applied to routine clinical data in a multicenter European memory clinic network achieved 78% accuracy in predicting AD diagnosis confirmed by subsequent biomarker assessment, with positive predictive value of 82%, suggesting clinical utility in stratifying patients for confirmatory biomarker testing [3, 15].

Challenges and Limitations

Despite remarkable technical progress, numerous challenges impede clinical translation of AI diagnostic tools for neurodegenerative diseases. Dataset limitations represent a fundamental constraint, as most models are trained on highly selected research cohorts with restricted demographic diversity, limiting generalizability to the broader clinical population and introducing risks of algorithmic bias against underrepresented ethnic and socioeconomic groups [1, 9, 16]. Prospective external validation in independent, diverse clinical cohorts remains the exception rather than the rule, and reported performance metrics may not reflect real-world diagnostic accuracy [14, 15].

The interpretability of complex deep learning models presents a significant barrier to clinical adoption. Clinicians and regulators require transparent, explainable AI systems that provide comprehensible rationales for diagnostic recommendations, enabling meaningful human oversight and facilitating trust [6, 16]. Explainable AI methodologies, including saliency maps, SHAP values, and attention visualization, partially address this need but do not fully resolve the opacity of high-dimensional deep learning architectures [9, 13].

Ethical concerns include data privacy and security in handling sensitive neurological and genetic information, the potential for AI-generated diagnoses to cause psychological harm if communicated without appropriate clinical context, and questions of consent and data ownership in large-scale biobank and federated learning frameworks [1, 6]. Regulatory pathways for AI medical devices vary significantly across jurisdictions, creating uncertainty for developers and

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healthcare institutions [16]. The risk of over-reliance on AI recommendations and deskilling of clinical expertise represents an additional concern requiring careful management through appropriate training and workflow design [14].

Discussion

This review highlights the substantial and rapidly advancing evidence base supporting AI as a transformative tool for early diagnosis of neurodegenerative diseases. The convergence of large-scale biobanks, multimodal data acquisition, advanced deep learning architectures, and digital health technologies has created unprecedented opportunities to detect neurodegeneration at presymptomatic or prodromal stages, when therapeutic intervention may be most efficacious [3, 8, 13].

A key insight emerging from the literature is the superiority of multimodal over unimodal AI approaches, consistent with the multifactorial and heterogeneous biology of neurodegenerative diseases [9, 14, 16]. No single biomarker or data modality captures the full complexity of AD, PD, or ALS pathology, and integrative AI frameworks that synthesize neuroimaging, fluid biomarkers, genetic, clinical, and digital data consistently achieve higher diagnostic accuracy. This multimodal paradigm aligns with the National Institute on Aging and Alzheimer's Association biological definition of AD, which requires evidence of amyloid and tau pathology alongside neurodegeneration markers [5].

The emergence of minimally invasive and non-invasive biomarkers, particularly blood-based Abeta and phospho-tau assays and digital biomarkers from wearables and smartphones, is particularly significant for scalable population-level screening [2, 3, 11]. AI models capable of integrating these accessible data types with routine clinical information may enable cost-effective risk stratification, identifying individuals most likely to benefit from confirmatory PET or CSF biomarker testing. This tiered screening approach could substantially reduce the diagnostic burden on specialist services and accelerate enrolment in prevention trials [8, 15].

Federated learning represents a promising strategy for addressing dataset diversity limitations by enabling collaborative model training across geographically distributed institutions without sharing raw patient data, thereby preserving privacy while enhancing generalizability [1, 9]. Early federated learning studies in neuroimaging have demonstrated model performance comparable to centralized training, supporting feasibility of this approach for multicenter neurodegenerative disease AI research [16].

The path to clinical translation requires not only technical refinement but also rigorous prospective validation, regulatory approval, and evidence of clinical utility beyond diagnostic accuracy, including impact on patient management decisions, therapeutic outcomes, and health economic value [14, 15]. Interdisciplinary collaboration among neurologists, radiologists, data scientists, clinical trial specialists, patient advocates, ethicists, and regulators is essential to navigate these challenges and ensure that AI diagnostic tools reach patients equitably and responsibly [6, 16].

Conclusion

Artificial intelligence holds transformative potential for early diagnosis of neurodegenerative diseases, offering capacity to detect pathological changes years before clinical onset and to integrate complex multimodal data beyond human analytical capability. Evidence from large cohort studies and systematic reviews supports diagnostic accuracy exceeding conventional approaches for Alzheimer's disease, Parkinson's disease, and related conditions, with multimodal deep learning frameworks demonstrating the greatest performance.

Significant challenges remain in dataset diversity, prospective validation, interpretability, ethical governance, and clinical integration. Future research priorities include development and validation of federated learning platforms for diverse global cohorts, prospective clinical trials evaluating AI diagnostic impact on patient outcomes, advancement of explainable AI methodologies, and establishment of clear regulatory and ethical frameworks. The development of accessible, cost-effective, and clinically interpretable AI diagnostic tools will be essential for realizing the promise of early intervention in neurodegenerative disease and reducing the global burden of cognitive and motor disability.

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