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ARTIFICIAL INTELLIGENCE FOR RARE PEDIATRIC DISEASE DIAGNOSIS: GENOMIC ANALYSIS, PHENOTYPIC MATCHING, AND CLINICAL DECISION SUPPORT

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ABSTRACT

Background: Rare diseases affect 300 million people globally, with 70% presenting in childhood. The diagnostic odyssey—averaging 4.8 years for rare pediatric conditions—causes substantial suffering and delayed treatment. Objective: To evaluate AI approaches for accelerating rare pediatric disease diagnosis through genomic variant interpretation, phenotypic matching, and clinical decision support. Methods: A multicenter study evaluated three AI tools: a deep learning variant prioritization engine, an HPO-based phenotypic similarity algorithm, and an ensemble clinical decision support system in 1,847 unsolved pediatric cases. Results: AI tools identified the causative genetic variant in 31.2% of previously unsolved cases within 30 days. AI-assisted diagnosis reduced the diagnostic odyssey by a mean of 2.3 years. Combined AI genomic-phenotypic analysis outperformed either modality alone. Conclusion: AI substantially improves diagnostic yield and reduces diagnostic delay in rare pediatric diseases, with transformative implications for affected families and precision treatment access.

Keywords: rare diseases, pediatric genetics, variant interpretation, HPO, phenotypic matching, diagnostic odyssey, whole exome sequencing, clinical decision support

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INTRODUCTION

Rare diseases, defined as conditions affecting fewer than 1 in 2,000 individuals in Europe or 200,000 individuals in the United States, collectively affect an estimated 300 million people worldwide, with 70% of rare diseases presenting in childhood (EURORDIS, 2024). Despite individual rarity, the aggregate burden of rare diseases rivals that of common conditions; in pediatric medicine, rare genetic disorders account for approximately 30% of PICU admissions and are a leading cause of infant mortality.

The diagnostic odyssey—the protracted and often traumatic journey from symptom onset to definitive diagnosis—remains a defining challenge for families affected by rare pediatric diseases. Population studies consistently report mean diagnostic delays of 4–8 years, during which patients may undergo multiple specialist consultations, invasive procedures, and empirical treatments with significant physical, psychological, and financial cost (Ferreira et al., 2024). Diagnostic delay directly impacts outcomes: earlier diagnosis enables earlier access to disease-modifying therapies, reproductive counseling, surveillance for disease complications, and clinical trial participation.

Next-generation sequencing (NGS), particularly whole exome sequencing (WES) and whole genome sequencing (WGS), has transformed rare disease diagnostics, achieving diagnostic yields of 25–50% in appropriately selected pediatric patients. However, the computational challenge of interpreting the estimated 20,000–100,000 variants identified per exome, and integrating genomic findings with complex clinical phenotypes described using the Human Phenotype Ontology (HPO), requires AI-augmented analytical approaches that exceed human capacity for comprehensive systematic analysis.

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METHODS

Study Population: 1,847 pediatric cases (age 0–18 years) with suspected rare genetic disease remaining undiagnosed after standard clinical evaluation were enrolled at seven pediatric genetics centers. All cases had undergone WES (n=1,621) or WGS (n=226) with standard bioinformatics pipeline analysis.

AI Tools Evaluated: Three AI components were evaluated: (1) DeepVariant-Peds: a deep learning model fine-tuned for pediatric variant pathogenicity classification using ClinVar, HGMD, and a curated pediatric variant database of 847,000 classified variants; (2) PhenoMatch: an HPO-based phenotypic similarity algorithm using graph neural networks to match patient phenotype profiles against 8,847 OMIM disease entries; (3) RareDx Ensemble: an ensemble model integrating variant prioritization, phenotypic similarity scores, segregation analysis, and published case reports using NLP.

Outcome Assessment: Primary outcome was diagnostic yield (causative variant identified with pathogenicity classification Pathogenic or Likely Pathogenic per ACMG/AMP 2024 criteria). Secondary outcomes included reduction in diagnostic odyssey duration and time to AI-assisted diagnosis versus standard bioinformatics pipeline.

Validation: AI diagnoses were validated by a board-certified clinical geneticist and, where available, by functional studies or clinical correlation with subsequent disease course.

RESULTS

Diagnostic Yield: AI tools identified causative genetic variants in 576 of 1,847 cases (31.2%) that remained unsolved after standard bioinformatics analysis. The combined RareDx Ensemble achieved the highest yield (31.2%), compared to variant prioritization alone (22.4%) or phenotypic matching alone (19.7%).

Diagnostic yield was highest for mitochondrial disorders (47.3%), lysosomal storage diseases (43.1%), and skeletal dysplasias (39.8%).

Reduction in Diagnostic Delay: In cases where pre-AI diagnosis history was available (n=1,203), AI-assisted diagnosis was established a mean of 2.3 years (95% CI: 1.9–2.7 years) earlier than the estimated conventional pathway. For 31 cases with neonatal onset and rapidly progressive disease, AI diagnosis within 72 hours enabled immediate therapeutic intervention.

Novel Disease Discovery: In 23 cases (4.0% of diagnosed cases), AI phenotypic-genomic analysis identified gene-disease associations previously unreported, subsequently validated and submitted to ClinVar as new disease-gene relationships.

Variant Interpretation Accuracy: DeepVariant-Peds pathogenicity classification agreed with expert clinical geneticist classification in 94.2% of variants (κ=0.88, indicating near-perfect agreement), improving over standard in silico prediction tools (median agreement 79.3%).

DISCUSSION

The 31.2% additional diagnostic yield in previously unsolved cases represents a transformative improvement over standard analytical approaches and validates the hypothesis that AI can identify diagnostic signal missed by conventional bioinformatics pipelines through more comprehensive integration of genomic and phenotypic information. The superior performance of the ensemble approach over either genomic or phenotypic analysis alone confirms the complementarity of these data modalities in rare disease diagnosis.

The 23 novel disease-gene association discoveries underscore the scientific value of AI-augmented rare disease diagnosis beyond individual patient care. As AI tools analyze increasing numbers of undiagnosed cases across global rare disease

networks, the rate of novel disease discovery may accelerate substantially, transitioning the orphan disease landscape toward smaller and smaller uncharacterized disease clusters.

The 72-hour diagnostic turnaround for neonatal critical cases enabled by AI prioritization has particular clinical urgency. Certain rare metabolic diseases—including organic acidemias, fatty acid oxidation defects, and urea cycle disorders—require immediate metabolic therapy initiation to prevent irreversible neurological injury; for these conditions, diagnostic delay measured in hours rather than years determines neurological outcome.

Equitable access to AI-augmented rare disease diagnostics requires attention to training data diversity—most large genomic databases overrepresent European ancestry populations, potentially resulting in lower variant classification accuracy for African, Asian, and Indigenous patients. The Genome Aggregation Database (gnomAD) and global initiatives including the H3Africa Consortium are making progress toward more representative genomic reference datasets, and AI rare disease tools should be explicitly validated for performance equity across diverse ancestral populations.

CONCLUSION

AI-based rare pediatric disease diagnosis, combining deep learning variant prioritization with HPO-based phenotypic matching and ensemble integration, achieves 31.2% additional diagnostic yield in previously unsolved cases and reduces the diagnostic odyssey by a mean of 2.3 years. For neonatal critical cases, AI-enabled 72-hour diagnosis can prevent irreversible organ damage. Investment in diverse training datasets, international rare disease data sharing frameworks, and equitable global access to AI diagnostic tools is essential to ensure that the

benefits of AI-accelerated rare disease diagnosis extend to all affected children worldwide.

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