RESEARCH ARTICLE

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Artificial intelligence and rare cardiovascular diseases: New frontiers in early diagnosis and prognostication

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ABSTRACT Rare cardiovascular diseases (RCDs) represent a diverse group of conditions that, although individually uncommon, collectively contribute to significant morbidity and mortality. Their diagnosis is often delayed due to heterogeneous presentations, lack of awareness, and limited access to specialized care—challenges that are particularly pronounced in India. Artificial intelligence (AI), encompassing machine learning, deep learning, and natural language processing, has emerged as a transformative tool in cardiovascular medicine, capable of detecting subtle patterns beyond human capacity and integrating multimodal data to enhance early diagnosis and prognostication. This review synthesizes global and India-specific evidence on the application of AI in RCDs. We highlight advances in imaging-based diagnostics, where AI-driven echocardiography, cardiac MRI, and CT angiography improve anomaly detection in hypertrophic cardiomyopathy and congenital heart disease. Genomic and biomarker-based approaches demonstrate how AI can accelerate variant classification and risk prediction in inherited cardiomyopathies and channelopathies. Clinical data integration using electronic health records and natural language processing further enables identification of rare phenotypes and supports decision-making. Beyond diagnosis, AI offers significant promise in prognostication and risk stratification, improving sudden cardiac death prediction, guiding implantable cardioverter-defibrillator (ICD) implantation, and identifying candidates for advanced therapies such as transplantation or gene-based treatments. While challenges persist—including data scarcity, algorithmic bias, infrastructural limitations, and ethical concerns—India's expanding digital health ecosystem and policies such as the National Rare Disease Policy (2021) and Ayushman Bharat Digital Mission provide unique opportunities. We argue that India is well-positioned to harness AI for RCDs by building multicentric datasets, developing AI-integrated rare disease registries, and fostering multidisciplinary collaboration. With public–private partnerships and global cooperation, India can transform the care of rare cardiovascular diseases, setting a precedent for other resource-limited settings.

KEYWORDS artificial intelligence, machine learning, rare cardiovascular diseases, hypertrophic cardiomyopathy, channelopathies, prognostication, precision cardiology, digital health, risk stratification

1. INTRODUCTION

1.1 Burden of cardiovascular diseases globally and in India

Cardiovascular diseases (CVDs) remain the leading cause of morbidity and mortality worldwide, accounting for an estimated 17.9 million deaths annually. While much attention has traditionally focused on common conditions such as ischemic heart disease, hypertension, and heart failure, rare cardiovascular diseases (RCDs) represent an underrecognized yet clinically significant subset. These conditions, though individually uncommon, collectively contribute to a considerable health burden due to diagnostic complexity, delayed treatment, and poor prognostic outcomes [1]–[3].

India, home to over 1.4 billion people, is undergoing an epidemiological transition where non-communicable diseases now account for the majority of deaths. Cardiovascular conditions dominate this trend, with India shouldering nearly one-fifth of the global CVD burden. Within this vast landscape, RCDs are often neglected due to limited awareness, lack of registry data, and diagnostic delays. Their true prevalence remains underestimated, yet anecdotal reports and hospital-based studies suggest that India harbors a substantial number of patients suffering from congenital, genetic, and rare structural heart diseases [4], [5].

1.2 Why Rare cardiovascular diseases (rcds) matter: Epidemiology, delayed diagnosis, prognosis challenges

RCDs encompass a heterogeneous group, including hypertrophic cardiomyopathy, arrhythmogenic right ventricular cardiomyopathy, congenital structural malformations, channelopathies, and inflammatory vasculitides such as Takayasu arteritis. These conditions often present with atypical or subtle symptoms, making early diagnosis challenging. Many patients endure years of diagnostic odysseys, during which disease progression worsens outcomes and limits therapeutic options [6], [7].

Prognostication poses another challenge. Rare cardiomyopathies, for example, may predispose young patients to sudden cardiac death, yet conventional risk models are poorly validated for these populations. The rarity of cases limits large-scale clinical trials, leading to gaps in evidence-based management. Consequently, patients frequently experience uncertainty, delayed intervention, and poorer survival outcomes compared to those with more common cardiovascular conditions [8], [9].

1.3 Emergence of AI as a transformative tool in cardiology

The rise of artificial intelligence (AI) has opened new frontiers in cardiovascular medicine. Machine learning (ML) and deep learning (DL) algorithms excel at detecting subtle patterns within large, complex datasets—an ability particularly valuable for RCDs, where human interpretation often falls short. AI has demonstrated utility in automated imaging interpretation, genomic analysis, predictive modeling, and integration of multimodal clinical data. These tools not only accelerate diagnosis but also hold promise for individualized risk stratification and prognostication [10]–[13].

In cardiology, AI applications have already shown success in arrhythmia detection, heart failure prediction, and coronary imaging. Extending these innovations to RCDs offers the possibility of reducing diagnostic delays, improving prognostic accuracy, and tailoring treatments to patient-specific profiles.

1.4 Rationale for focusing on india (demographics, health system challenges, digital health ecosystem)

India represents a unique context for studying AI applications in RCDs. The country's sheer population size, genetic diversity, and burden of undiagnosed cases create both opportunities and challenges. Healthcare access remains uneven, with advanced diagnostic facilities concentrated in urban tertiary hospitals, leaving rural populations underserved. Awareness of RCDs among primary care providers is limited, contributing to diagnostic delays.

At the same time, India is rapidly expanding its digital health ecosystem. Initiatives such as the Ayushman Bharat Digital Mission aim to create interoperable health records, while the growth of telemedicine and mobile health platforms has increased digital connectivity. These developments provide fertile ground for deploying AI solutions that can bridge diagnostic gaps, particularly in resource-limited settings.

2. OBJECTIVES OF THE REVIEW

This review aims to:

1. Provide an overview of rare cardiovascular diseases and their current burden in India.
2. Examine the role of artificial intelligence in enhancing early diagnosis and prognostication of RCDs.
3. Highlight India-specific challenges and opportunities for implementing AI-driven solutions.
4. Identify research gaps and propose directions for future collaboration across medicine, technology, and policy.

3. RARE CARDIOVASCULAR DISEASES: AN OVERVIEW

3.1 Definition and classification

Rare cardiovascular diseases (RCDs) are a diverse group of conditions that occur with relatively low prevalence but often carry disproportionate morbidity and mortality. The classification of RCDs typically includes:

- **Congenital diseases**: structural heart defects such as single-ventricle physiology or anomalous coronary artery origins.
- **Genetic cardiomyopathies**: hypertrophic cardiomyopathy (HCM), dilated cardiomyopathy of genetic origin, arrhythmogenic cardiomyopathy.
- **Inherited channelopathies**: long QT syndrome, Brugada syndrome, catecholaminergic polymorphic ventricular tachycardia (CPVT).
- **Inflammatory and autoimmune conditions**: Takayasu arteritis (more prevalent in India), Kawasaki disease, myocarditis of rare etiologies.
- **Metabolic and storage disorders**: Fabry disease, mitochondrial cardiomyopathies.
- **Structural anomalies and vascular malformations**: rare aortic and pulmonary vascular disorders.

Although each disease is uncommon in isolation, together they represent a meaningful burden on healthcare systems due to diagnostic complexity, specialized care requirements, and high risk of adverse outcomes.

3.2 Epidemiology and prevalence in India

Estimating the prevalence of RCDs in India is challenging due to limited registry data, underdiagnosis, and fragmented reporting systems. Congenital heart disease (CHD) is one of the better-documented conditions, with estimates suggesting an incidence of 8–10 per 1,000 live births, translating into nearly 200,000 new cases annually. Among genetic cardiomyopathies, hypertrophic cardiomyopathy is thought to affect 1 in 500 individuals globally, yet its prevalence in India remains underexplored due to diagnostic limitations [10]–[13].

Certain RCDs exhibit geographic clustering in India. For example, Takayasu arteritis, a rare large-vessel vasculitis, is more commonly reported among Indian and East Asian populations. Similarly, case series highlight delayed recognition of inherited arrhythmia syndromes, with many patients presenting only after adverse cardiac events. The lack of systematic screening and specialized diagnostic centers means that the actual burden is likely far higher than current estimates suggest.

3.3 Clinical and diagnostic challenges

The diagnosis of RCDs is fraught with challenges:

- **Clinical heterogeneity**: Symptoms often mimic common conditions such as asthma, anemia, or routine arrhythmias, leading to misdiagnosis.

- **Lack of awareness**: Primary care physicians and even general cardiologists may have limited exposure to rare phenotypes, delaying referral to specialized centers.
- **Resource limitations**: Advanced diagnostic modalities such as cardiac MRI, genetic sequencing, and electrophysiological mapping are not uniformly available across India.
- **Limited specialist centers**: Dedicated rare disease clinics and registries are scarce, resulting in fragmented care.
- **Socioeconomic barriers**: High out-of-pocket costs for advanced investigations deter timely diagnosis for many families.

Collectively, these challenges highlight the urgent need for novel approaches such as AI-driven tools that can identify subtle patterns across imaging, genomics, and electronic health records, thereby improving diagnostic precision and timeliness.

4. ARTIFICIAL INTELLIGENCE IN CARDIOVASCULAR MEDICINE

4.1 Definitions: AI, machine learning (ML), deep learning (DL), and natural language processing (NLP)

Artificial intelligence (AI) refers to the capability of computer systems to simulate human-like cognitive functions such as learning, reasoning, and decision-making. Within AI, machine learning (ML) denotes algorithms that identify patterns in data and improve performance through iterative training. Deep learning (DL) is a subset of ML that uses artificial neural networks with multiple layers to recognize complex, nonlinear patterns, particularly in large-scale imaging and genomic datasets. Natural language processing (NLP), another branch of AI, focuses on analyzing unstructured textual data, such as electronic health records (EHRs), medical literature, or patient narratives, and extracting clinically meaningful information [14]–[17].

These methods collectively form the foundation for AI applications in cardiovascular medicine, enabling automated image interpretation, predictive analytics, and data-driven decision support.

4.2 Evolution of AI applications in mainstream cardiology

AI has already revolutionized several domains of mainstream cardiology:

- **Arrhythmias**: AI-driven electrocardiogram (ECG) analysis enables early detection of atrial fibrillation, ventricular arrhythmias, and conduction abnormalities, often before they are clinically evident. Algorithms have been validated to recognize subtle ECG features that even expert cardiologists may overlook.
- **Heart Failure**: Predictive ML models integrate imaging, biomarkers, and EHR data to identify patients at risk of decompensation, guide therapy adjustments, and optimize outcomes.
- **Coronary Artery Disease**: Deep learning applied to coronary CT angiography (CCTA) can quantify plaque burden, characterize morphology, and predict future cardiac events with remarkable accuracy. Additionally, AI has been integrated into non-invasive functional testing (e.g., CT-derived fractional flow reserve). These advances highlight the potential of AI to transform cardiovascular care by improving precision, efficiency, and accessibility.

4.3 Why rare cardiovascular diseases (RCDs) need AI

The need for AI is arguably even greater in rare cardiovascular diseases (RCDs) due to three critical factors:

1. **Complexity**: RCDs often involve heterogeneous presentations that defy conventional diagnostic algorithms.
2. **Rarity**: The low prevalence makes it difficult for physicians to accumulate sufficient clinical experience, leading to misdiagnosis or delayed recognition.
3. **Pattern Recognition Beyond Human Capacity**: AI excels at identifying subtle, multidimensional patterns across imaging, genomics, and clinical data that may be invisible to human interpretation.

For instance, AI algorithms trained on multimodal data can differentiate between hypertrophic cardiomyopathy and athlete's heart, or flag subtle congenital anomalies on imaging studies. By leveraging large datasets and computational power, AI has the potential to reduce diagnostic delays, improve prognostication, and optimize resource allocation for RCDs.

5. AI IN THE DIAGNOSIS OF RARE CARDIOVASCULAR DISEASES

5.1 Imaging-based applications

5.1.1. Echocardiography, cardiac MRI, and CT angiography

Imaging is the cornerstone of diagnosing RCDs, yet interpretation often demands advanced expertise. AI-driven models can enhance detection accuracy, automate measurements, and reduce inter-observer variability.

- **Echocardiography**: DL algorithms can automatically quantify chamber size, wall thickness, and ejection fraction. For hypertrophic cardiomyopathy (HCM), AI can highlight regional wall motion abnormalities or subtle hypertrophy patterns.
- **Cardiac MRI (CMR)**: AI-based segmentation tools streamline the analysis of myocardial fibrosis and scar burden, essential for risk stratification in genetic cardiomyopathies.
- **CT Angiography**: In congenital heart disease, AI can reconstruct three-dimensional models, assisting in early recognition of complex vascular anomalies and aiding surgical planning.

5.1.2. Examples: hypertrophic cardiomyopathy and congenital heart disease

Studies have demonstrated that AI models outperform traditional diagnostic approaches in distinguishing HCM from hypertensive heart disease or athlete's heart—a diagnostic dilemma in young populations. Similarly, AI-assisted CMR has improved detection of congenital anomalies such as anomalous pulmonary venous return or coronary artery variants, which often escape early recognition [17]–[21].

5.2 Genomic and biomarker-based diagnostics

5.2.1. Integration of AI with next-generation sequencing (NGS)

Genomics is critical for diagnosing inherited cardiomyopathies and channelopathies. However, interpreting vast amounts of sequencing data is challenging. AI facilitates variant classification, pathogenicity prediction, and genotype-phenotype correlation, enabling faster and more accurate diagnoses [22], [23].

5.2.2. Predictive algorithms for genetic cardiomyopathies and channelopathies

For conditions like long QT syndrome or Brugada syndrome, AI-driven predictive models can integrate genomic data with ECG features to improve diagnostic specificity. Similarly, AI has been applied to stratify sudden cardiac death risk in hypertrophic cardiomyopathy by integrating genetic, imaging, and clinical variables [24], [25].

5.2.3. Use in India: Accessibility, affordability, and digital genomics initiatives

In India, genomic testing remains limited by high cost, lack of insurance coverage, and concentration in urban tertiary centers. However, initiatives such as the GenomeIndia Project and emerging collaborations between biotech companies and academic institutions are building large-scale genomic datasets. Integrating AI into these initiatives could make genomic diagnostics more scalable, affordable, and accessible, particularly for rare diseases.

5.3 Clinical data integration

5.3.1. Electronic health records (EHRs) and clinical decision support systems

EHRs contain valuable longitudinal data but are often fragmented. AI models can extract and harmonize data from EHRs to identify rare disease phenotypes. For instance, subtle patterns of recurrent unexplained syncope, family history of sudden cardiac death, and abnormal ECG findings could trigger alerts for inherited arrhythmia syndromes [26], [27].

5.3.2. NLP-based extraction of rare disease phenotypes

Natural language processing (NLP) enables AI to parse unstructured clinical notes, discharge summaries, and radiology reports. In rare cardiovascular diseases, NLP has been applied to identify patients with conditions such as arrhythmogenic right ventricular cardiomyopathy (ARVC) by recognizing textual patterns describing ventricular arrhythmias and imaging abnormalities [28], [29].

5.3.3. Case examples of AI-assisted rare cardiac syndrome identification

- In international studies, AI-enabled ECG interpretation has improved recognition of Brugada syndrome patterns that are often missed by human readers.
- Pilot projects in India have begun exploring AI-based ECG screening in rural health camps, with potential to flag high-risk cases for referral to tertiary centers.
- Combining EHR integration, imaging, and genomics, AI can create clinical decision support systems that assist cardiologists in managing complex RCD cases more effectively.

6. AI IN PROGNOSTICATION AND RISK STRATIFICATION

6.0.1. Machine learning models for predicting outcomes in RCDs

Rare cardiovascular diseases (RCDs) often pose significant prognostic uncertainty due to their heterogeneity and lack of large-scale clinical trials. Machine learning (ML) models can integrate diverse datasets—clinical variables, imaging features, genetic markers, and biochemical parameters—to generate individualized predictions of disease progression and outcomes. Unlike conventional risk scores, which are often developed for common conditions, AI-driven models adapt to complex, non-linear relationships within rare disease cohorts.

For example, ML algorithms have been trained to predict adverse ventricular remodeling in dilated cardiomyopathy or arrhythmic events in hypertrophic cardiomyopathy (HCM), often outperforming traditional scoring systems. Such predictive tools enable clinicians to identify high-risk patients earlier, facilitating timely interventions [30], [31].

6.0.2. Personalized medicine approaches

Personalized or precision medicine relies on tailoring treatment to the unique risk profile of an individual patient. AI has particular relevance here. In inherited cardiomyopathies and channelopathies, ML-based algorithms can combine genetic data with imaging and ECG features to estimate sudden cardiac death risk, a key determinant in decisions about implantable cardioverter-defibrillator (ICD) therapy [32], [33].

In HCM, for instance, AI-driven models integrating fibrosis burden from cardiac MRI, family history, and genotype data have demonstrated superior accuracy compared to guideline-based risk calculators. Such tools help refine management strategies, minimizing both under-treatment and unnecessary device implantation.

6.0.3. AI in identifying candidates for advanced therapies (ICDs, Transplant, Gene Therapy)

Beyond standard therapies, AI can guide advanced interventions:

- **ICDs**: By stratifying arrhythmic risk, AI ensures that ICD implantation is targeted to those most likely to benefit, thereby improving outcomes and optimizing resource utilization.
- **Transplantation**: AI-based prognostic models can predict progression to end-stage heart failure in rare cardiomyopathies, guiding timely referral for transplantation before irreversible deterioration occurs.
- **Gene Therapy**: In genetic disorders such as Fabry disease or Duchenne-related cardiomyopathy, AI algorithms can

help identify patients with specific mutational patterns who are most likely to benefit from emerging gene-based therapies.

6.0.4. Indian experience: Gaps and opportunities

In India, the application of AI for prognostication in RCDs remains limited but promising. Pilot projects have explored AI-assisted ECG interpretation for arrhythmia risk stratification, and some tertiary centers are experimenting with ML-based models for HCM risk prediction. However, several gaps persist:

- **Lack of large annotated datasets**: Without robust national registries, algorithm training is constrained.
- **Urban–rural disparity**: Most AI-enabled tools are deployed in tertiary centers, leaving vast rural populations underserved.
- **Cost barriers**: Access to advanced diagnostics such as MRI or genetic sequencing—often integral to AI models—remains limited for many families.

Opportunities lie in leveraging India's rapidly expanding digital health infrastructure, integrating AI with initiatives such as the Ayushman Bharat Digital Mission to build inclusive, large-scale datasets for rare cardiovascular conditions.

7. POTENTIAL BENEFITS AND OPPORTUNITIES

7.1 Earlier detection and reduced diagnostic delay

AI offers the promise of shortening the "diagnostic odyssey" that many RCD patients face. By flagging subtle imaging anomalies, recognizing rare ECG patterns, or extracting phenotypic cues from electronic records, AI can prompt earlier diagnosis compared to conventional methods. In India, where awareness of RCDs is low and delays are common, early detection could significantly reduce morbidity and mortality.

7.2 Improved risk stratification, clinical decision-making, and personalized care

AI-driven prognostic models enable granular risk stratification, moving beyond population averages to individualized predictions. This empowers clinicians to make better-informed decisions, whether in initiating preventive therapies, selecting surgical interventions, or tailoring follow-up schedules. For adolescents and young adults with inherited cardiomyopathies, personalized care informed by AI could mean the difference between a normal lifespan and premature mortality.34,35

7.3 Cost-effectiveness in resource-limited Indian settings

One of the most compelling arguments for AI in India is its potential to improve cost-effectiveness. Traditional diagnostic modalities for RCDs—such as cardiac MRI, genetic testing, or invasive electrophysiology—are expensive and often inaccessible outside metropolitan centers. AI tools, once developed, can scale at relatively low incremental cost, especially when integrated with widely available technologies like echocardiography and ECG. Cloud-based AI platforms and mobile-enabled diagnostic support can bring expert-level interpretation to remote areas, thereby democratizing access to advanced cardiovascular care.36,37

7.4 Potential to strengthen national rare disease registries and precision health initiatives

India's National Rare Disease Policy (2021) emphasizes the creation of registries and databases. AI can serve as both a data aggregator and analytic engine, standardizing inputs from multiple centers, identifying hidden patterns, and continuously updating risk prediction models. Such registries would not only improve epidemiological understanding but also accelerate clinical research and policy planning [38].

Furthermore, integrating AI with precision health initiatives could allow India to leapfrog traditional barriers, creating a digital ecosystem where rare cardiovascular diseases are systematically diagnosed, monitored, and treated with evidence-based strategies.

8. CHALLENGES AND LIMITATIONS

8.1 Data scarcity and imbalance in RCDs

A fundamental challenge in applying AI to rare cardiovascular diseases (RCDs) is the scarcity of high-quality datasets. By definition, RCDs affect small populations, limiting the number of cases available for algorithm training. This scarcity often leads to imbalanced datasets, where the minority class (rare disease cases) is overshadowed by more common cardiovascular conditions. Such imbalance increases the risk of model overfitting and reduces accuracy when applied to real-world scenarios. In India, where centralized rare disease registries are still in their infancy, the lack of comprehensive data further hinders the development of robust AI tools.

8.2 Algorithmic bias and generalizability issues (Global vs. Indian Cohorts)

Most AI models in cardiology are developed using datasets from high-income countries. These algorithms may not generalize well to Indian populations due to differences in genetics, environmental exposures, comorbidities, and healthcare delivery systems. For example, risk prediction tools validated in Western cohorts may underperform when applied to Indian patients with unique epidemiological characteristics, such as higher prevalence of rheumatic heart disease or earlier onset of cardiomyopathies. Without context-specific validation, AI risks perpetuating bias and widening disparities in diagnosis and care.

8.3 Infrastructure limitations (Cloud Computing, Interoperability of EHRs)

AI applications require significant computational power, reliable internet connectivity, and seamless integration across healthcare systems. In India, many hospitals still rely on fragmented record-keeping systems, with limited adoption of electronic health records (EHRs). Even where EHRs exist, interoperability across institutions is poor, restricting the aggregation of large-scale datasets necessary for training AI algorithms. Cloud computing infrastructure, while expanding, is not universally available in rural and semi-urban areas, further constraining AI deployment outside major tertiary centers.

8.4 Ethical and legal considerations: data privacy, consent, accountability

The deployment of AI in healthcare raises critical ethical and legal concerns. Protecting patient data privacy is paramount, especially when sensitive genomic and cardiovascular data are involved. Informed consent processes for AI-enabled tools are often inadequately defined, leaving patients unaware of how their data may be used. Furthermore, accountability in AI-driven decision-making remains unresolved—if an AI algorithm misclassifies a rare disease, who bears responsibility: the physician, the hospital, or the developer? India currently lacks comprehensive legal frameworks that clearly address these concerns, underscoring the need for robust data governance policies.

8.5 Cost and accessibility challenges in low-resource Indian healthcare systems

Although AI holds the potential to reduce costs in the long term, the initial investment in hardware, software, and training can be prohibitive. Many Indian public hospitals operate under constrained budgets, prioritizing essential services over advanced technology. Patients from low-income backgrounds face additional barriers, as they may not afford the diagnostic tests (e.g., cardiac MRI, genetic sequencing) upon which many AI models depend. Without affordable, scalable solutions, AI risks being restricted to elite centers, exacerbating healthcare inequities.

9. THE INDIAN CONTEXT: OPPORTUNITIES AND BARRIERS

9.1 Healthcare infrastructure

India's healthcare system is a patchwork of advanced tertiary care facilities alongside resource-limited primary and secondary centers. Tertiary hospitals in metropolitan cities such as Delhi, Mumbai, and Chennai are equipped with specialized cardiology units and advanced imaging technologies capable of supporting AI integration. However, access is uneven—urban–rural disparities remain stark, with rural populations often deprived of advanced diagnostics and specialized expertise. This uneven distribution represents both a barrier and an opportunity: AI-enabled tools could potentially extend the reach of expert-level diagnostics to underserved areas via cloud-based and telemedicine platforms [39].

9.2 Digital health ecosystem

The Indian government has made significant strides in digital health. The Ayushman Bharat Digital Mission (ABDM) seeks to create a unified digital health infrastructure by linking patient records, providers, and healthcare facilities. E-health initiatives and telecardiology programs are expanding access to cardiovascular care, especially in remote regions. AI integration with these platforms could allow real-time screening and triaging of RCDs, ensuring early referrals to specialized centers. By leveraging India's mobile penetration and growing internet coverage, AI can transform rural cardiovascular care into a digitally inclusive ecosystem [40].

9.3 Policy and regulatory framework

The National Rare Disease Policy (2021) acknowledges the importance of rare disease registries and the need for innovative diagnostic approaches. While AI is not explicitly integrated into this policy, the framework provides a foundation for incorporating digital health and AI solutions. Moreover, the rise of India's data protection laws and guidelines on responsible AI use provides an opportunity to establish ethical frameworks tailored to healthcare. Ensuring that AI in RCDs aligns with these policies will be crucial for building trust, safeguarding patient rights, and scaling adoption.

9.4 Research and capacity building

AI in cardiology research is still in its nascent stages in India, with isolated pilot projects demonstrating feasibility in areas such as ECG interpretation and echocardiography automation. However, large-scale, multicenter studies on RCDs are scarce. Building capacity requires cross-disciplinary collaboration, training cardiologists to understand AI technologies and data scientists to appreciate clinical contexts. Academic partnerships, public–private collaborations, and government funding can accelerate this process. Over time, India has the potential to emerge as a leader in AI-driven rare disease research, leveraging its vast and diverse population as a unique data resource.

10. FUTURE DIRECTIONS

10.1 Need for large-scale, multicentric datasets in India

The success of AI in rare cardiovascular diseases (RCDs) hinges on access to large, high-quality datasets. In India, data on RCDs are fragmented across tertiary centers, with limited standardization or central aggregation. Moving forward, there is an urgent need for multicentric data networks that capture clinical, imaging, and genomic information from diverse populations. Such initiatives would reduce algorithmic bias, enhance generalizability, and enable more accurate predictions tailored to Indian cohorts.

10.2 Development of rare cardiovascular disease registries integrated with AI tools

A critical step toward harnessing AI is the establishment of dedicated national RCD registries. These registries, integrated with AI analytics, could automate disease surveillance, detect emerging trends, and provide real-time insights into patient outcomes. For example, AI-driven algorithms could continuously scan registry data to identify clusters of undiagnosed cardiomyopathies or predict progression to advanced heart failure. Linking registries with the Ayushman Bharat

Digital Mission would further expand reach and interoperability.

10.3 Multidisciplinary collaboration between specialists

RCDs sit at the intersection of multiple disciplines—cardiology, genetics, bioinformatics, data science, and public health. Effective use of AI requires cross-disciplinary collaboration, where cardiologists provide clinical insights, geneticists contribute molecular understanding, and AI specialists build robust computational frameworks. India should invest in training programs and collaborative platforms that encourage this synergy, thereby building a workforce capable of driving innovation in AI-enabled cardiovascular care.

10.4 Role of public–private partnerships in building AI-enabled healthcare solutions

Given the scale and complexity of India's healthcare needs, public–private partnerships (PPPs) will be essential. Collaborations between government agencies, academic institutions, technology firms, and healthcare providers can accelerate the development and deployment of AI solutions. Industry partners can provide computational infrastructure and technical expertise, while public institutions ensure equity, regulatory oversight, and ethical safeguards. PPPs also hold potential to make AI solutions more affordable and scalable, ensuring that innovations benefit not just urban elite centers but also rural and underserved populations.

10.5 Global collaborations with adaptation for Indian populations

Finally, India should actively participate in global collaborations on AI in cardiology, leveraging international datasets, algorithms, and best practices. At the same time, these global solutions must be contextually adapted to reflect India's unique genetic diversity, epidemiological trends, and resource constraints. By contributing its large and heterogeneous population data, India can not only benefit from global knowledge but also play a leading role in shaping the future of AI-driven cardiovascular medicine.

11. CONCLUSION

Artificial intelligence represents a paradigm shift in the diagnosis and prognostication of rare cardiovascular diseases. By leveraging imaging, genomics, and clinical data, AI has the capacity to overcome traditional barriers of rarity, complexity, and delayed recognition. Early studies demonstrate that AI can enhance risk stratification, guide personalized therapy, and improve patient outcomes, offering a new era of precision medicine in cardiology.

Yet, this promise must be balanced against limitations. Data scarcity, algorithmic bias, infrastructural constraints, and ethical concerns remain significant hurdles, particularly in resource-constrained healthcare systems such as India's. Without careful regulation, inclusive data collection, and context-specific validation, AI risks deepening existing inequities rather than alleviating them.

India, however, is uniquely positioned to lead this transformation. With its large population base, growing digital health infrastructure, and emerging innovation ecosystem, the country has an unparalleled opportunity to combine big data with AI innovation for rare cardiovascular disease care. National initiatives like the Ayushman Bharat Digital Mission and the National Rare Disease Policy provide a framework within which AI-driven solutions can be embedded for maximum impact.

This review calls for collaborative action across stakeholders:

- Clinicians must engage in building and validating AI tools that reflect clinical realities.
- Researchers and data scientists must co-develop algorithms with cultural, genetic, and epidemiological relevance to Indian populations.
- Policymakers must establish ethical, legal, and regulatory frameworks that ensure safe, equitable use of AI.
- Technology developers and industry partners must work with healthcare providers to create scalable, affordable solutions.

By uniting these efforts, India can ensure that AI does not remain a futuristic concept but becomes a practical, inclusive, and transformative tool in the fight against rare cardiovascular diseases, reshaping patient care for generations to come.

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