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Transforming Rare Disease Diagnosis with AI

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Abstract

Artificial intelligence is changing healthcare fast. It is making diagnoses accurate, helping doctors get better results, and streamlining how care works. This paper looks at how AI shows up in healthcare right now – where it is already making a difference, what is working, and what is still tricky. The focus is on machine learning, natural language processing, and computer vision. Particular attention is given to using AI in diagnosing rare diseases by integrating genomic and phenotypic data, addressing the limitations of traditional diagnostic methods. Rare diseases are highly diverse and complex, which leads to major gaps in research, medical expertise, diagnosis, treatment options, and overall patient quality of life. Although each rare disease affects only a small fraction of the population – around 1 in 2,000 people in Europe and 1 in 1,250 in the United States – the total number of such conditions is huge, with over 5,000 identified worldwide. As a result, rare diseases collectively impact millions of people, affecting nearly 30 million individuals in Europe and about 25 million in North America. Rare diseases affect a small number of people but cause huge health and financial stress, especially in India. Because of genetic diversity and practices like consanguineous marriages, India faces extra challenges in identifying and managing these conditions. Although the National Policy for Rare Diseases (2025) aimed to improve care, awareness among doctors and the public is still low, diagnoses are often delayed, and treatments can cost over ₹1 crore while government support is limited. Primary care lacks early screening and strong referral systems, and access to affordable orphan drugs remains difficult. However, the recent Union Budget 2026–27 brought important relief by cutting import duties on costly cancer and rare disease medicines and proposing customs-duty exemptions for seven additional rare diseases – showing that the government is waking up to the needs of patients. To make care truly equitable, India still needs better early diagnosis, revised health technology assessment methods for rare diseases, more local drug production, and rare disease training in medical education. Advanced AI models like deep learning and clustering uncover novel disease patterns, refine diagnostics, and enable personalized medicine, addressing challenges in data and ethics.

Keywords: Artificial intelligence (AI), deep learning, disease prediction, genotype-phenotype integration, machine learning (ML), natural language processing (NLP), rare disease diagnosis

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INTRODUCTION

Artificial intelligence is shaking up many fields, but you really notice it in healthcare. Over the last few decades, AI has shown it can make a real difference – helping doctors make better decisions, cutting down on paperwork, and making things better for patients. From machine learning algorithms capable of diagnosing diseases from medical images to predictive analytics that help in early detection and personalized treatment plans to NLP systems designed to process medical documents, sentences, and individual words for extracting valuable information, AI is rapidly reshaping the healthcare landscape [1]. The rapid development of artificial intelligence (AI) is beginning to transform the landscape of rare disease research and clinical care.

ROLE OF AI IN HEALTHCARE

AI is changing healthcare in ways we could not have imagined a decade ago. It is not just about faster computers or smarter apps – AI actually helps doctors spot diseases earlier and with more accuracy, especially in tricky areas like cancer or neurological problems. Medical imaging? AI makes that whole process sharper and quicker. And when it comes to rare diseases, machine learning and deep learning are uncovering things we would probably miss on our own (Figure 1).

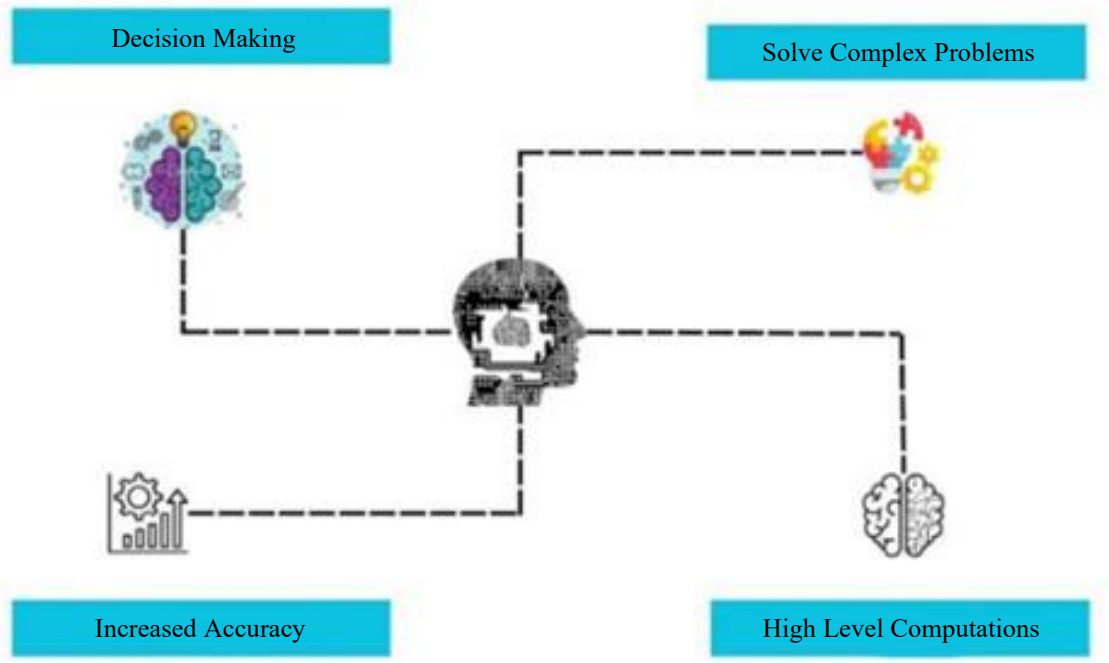

Figure 1. Major Roles of Artificial Intelligence in Healthcare.

On the admin side, AI takes care of a ton of boring work – scheduling appointments, handling billing, and smoothing out workflow issues – so people can focus on actual patient care. It is also shaking up drug discovery, speeding up clinical trials, and pushing personalized medicine forward by digging deep into patient data. All of this is not just about efficiency; it is making healthcare smarter and more personal. AI technologies are revolutionizing healthcare by integrating genotypic and phenotypic data, enabling the discovery of complex patterns that aid in diagnosing rare diseases. AI simplifies the analysis of complex genomic and phenotypic data, helping healthcare providers find new biomarkers and understand how genes relate to diseases [2–4].

Overview of Rare Diseases

AI is changing the way we tackle rare diseases. Machine learning and deep learning dig through mountains of medical data – genetic info, patient histories, all of it – and spot patterns nobody noticed before. This means doctors can diagnose people faster, come up with new treatment options, and even get a sense of how these diseases might progress. In the end, that is real hope for finding answers earlier and making diagnoses that actually stick [5].

Motivation for Research

Children's Hospital of Philadelphia (CHOP) has been a leader in integrating AI into rare disease phenotyping, which uses facial recognition AI to identify syndromes based on phenotypic features [6]. Another example is Northwestern University's Feinberg School of Medicine, which focuses on machine learning for deep phenotyping [7]. Despite its critical role in medical genetics, there is still a significant gap in the establishment of standardized protocols for phenotype assessment, which has led to variability in clinical applications and research outcomes. Several studies have emphasized the need for more structured frameworks to accurately interpret and utilize phenotypic information in clinical genetics.

METHODOLOGIES AND TECHNOLOGIES

Methodologies Involved

Research Objective

This study looks at how artificial intelligence can help diagnose and predict rare diseases by digging into medical data – everything from clinical records to imaging and even genetic info. The goal is to build an AI-powered system that catches rare diseases early and helps doctors tailor treatments to each patient [8].

Study Design

This study takes a quantitative approach, zeroing in on machine learning and deep learning techniques to dig into datasets on rare diseases [9]. The process goes like this: gather the data, build the AI models, train them, and then see how well they perform.

Data Collection

Data for this study were obtained from multiple sources:

- *Clinical Data:* Patient records, including diagnostic information, medical history, and clinical notes.
- *Medical Imaging Data:* X-rays, MRIs, and CT scans containing potential signs of rare diseases [10].
- *Genomic Data:* DNA sequencing and mutation data related to rare genetic disorders.
- *Public Datasets:* Rare disease databases, such as Orphanet and RARE were used to supplement the data.

Before diving in, the data were preprocessed – filled in missing info, normalized features, and converted any categories that needed it.

AI Model Development

AI models were developed using the following approaches:

- *Machine Learning Models:* Random Forest (RF) and Support Vector Machine (SVM) were employed for classification tasks, including predicting the presence of rare diseases based on clinical data [11].
- *Deep Learning Models:* Convolutional Neural Networks (CNNs) were used to classify medical images (e.g., MRI scans) for detecting visual patterns associated with rare diseases [12].

Recurrent Neural Networks (RNNs) were utilized to analyze genomic sequences for identifying mutations related to rare genetic conditions. Transfer learning was implemented using pretrained models (e.g., ResNet and VGGNet) for medical image analysis, which is particularly useful when training data for rare diseases is limited.

Model Training and Optimization

Models were trained using a training dataset comprising 80% of the total available data, with the remaining 20% reserved for testing. Hyperparameter optimization was conducted using grid search and random search techniques. Data augmentation – things like rotating and flipping medical images – were used to help the deep learning models handle a wider range of cases [13].

Technologies Involved

Machine Learning (ML)

ML involves training models on data to detect patterns and make predictions. ML can enhance diagnostic accuracy in rare genetic diseases. In ML algorithms, we use both supervised and unsupervised learning. Supervised learning is commonly used in genetic studies (86.7%), with Random Forest (RF) being the most popular method (73.7). RF works by combining many decision trees, making it good at handling complex data and identifying the most important features for predicting genetic problems. Unsupervised learning clusters phenotypic data to reveal hidden relationships between symptoms and rare diseases, aiding discovery without prior labels (Figure 2) [14].

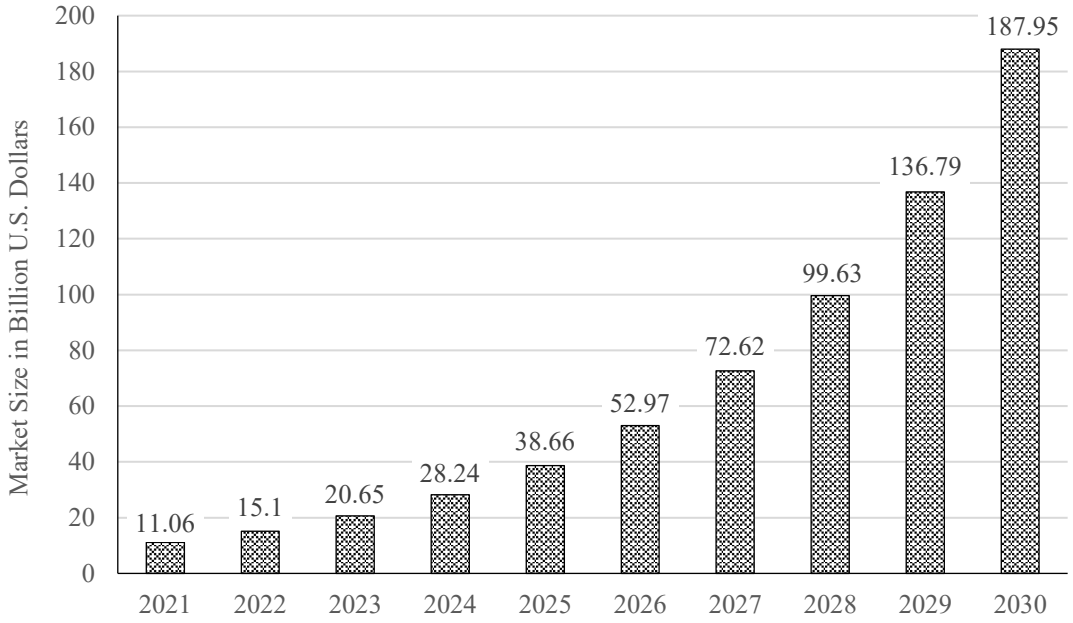

Figure 2. Market size of AI in healthcare (in US dollar).

Deep Learning (DL)

Deep learning leverages neural networks to interpret large, complex datasets. Here we come with the first deep learning approach SHEPHERD for diagnosis of rare genetic disease [15]. SHEPHERD is a diagnostic tool that uses knowledge of diseases, phenotypes, and genes to find new links between a patient's clinical and genomic data. It was evaluated on a dataset of 465 patients with 299 rare diseases. It identifies causal genes (average rank: 3.52), finds similar patients, and interprets novel disease presentations, aiding complex diagnoses in rare cases. It identifies causal genes (average rank: 3.52), finds similar patients, and interprets novel disease presentations, aiding complex diagnoses in rare cases.

Natural Language Processing (NLP)

NLP processes unstructured text data, such as electronic health records (EHRs), to extract relevant phenotypic and genomic information. It can spot rare diseases in all that clutter, find the key facts automatically, and save people from tons of tedious work. That is where Named Entity Recognition, or NER, comes in. NER hunts down and labels disease info buried in unstructured text. But it is not always easy. You must deal with a mess of synonyms (like "NF1" instead of "neurofibromatosis type I"), overlapping symptoms, and language that jumps between casual and super technical [16]. The old way relied on hand-written rules, but now, most people train models on huge piles of data. Still, even with all this tech, NER has not really been pushed far enough when it comes to rare diseases.

Genomic Sequencing and Phenotyping Models

Genomic sequencing and phenotyping are critical in understanding genetic diseases, especially rare conditions. Genomic sequencing reveals an individual's DNA, identifying mutations like single nucleotide polymorphisms (SNPs) and structural variations linked to diseases. It helps uncover genetic variants associated with specific health conditions. Phenotyping models use an individual's phenotypes to identify and rank candidate diagnostic genes.

These models are based on the idea that disease-related phenotypes are caused by one or more gene dysfunctions. Tools like Face2Gene and DeepGestalt have used phenotypic data, such as facial images, to identify rare genetic syndromes [17]. These models help bridge the gap between genotype and phenotype (Figure 3).

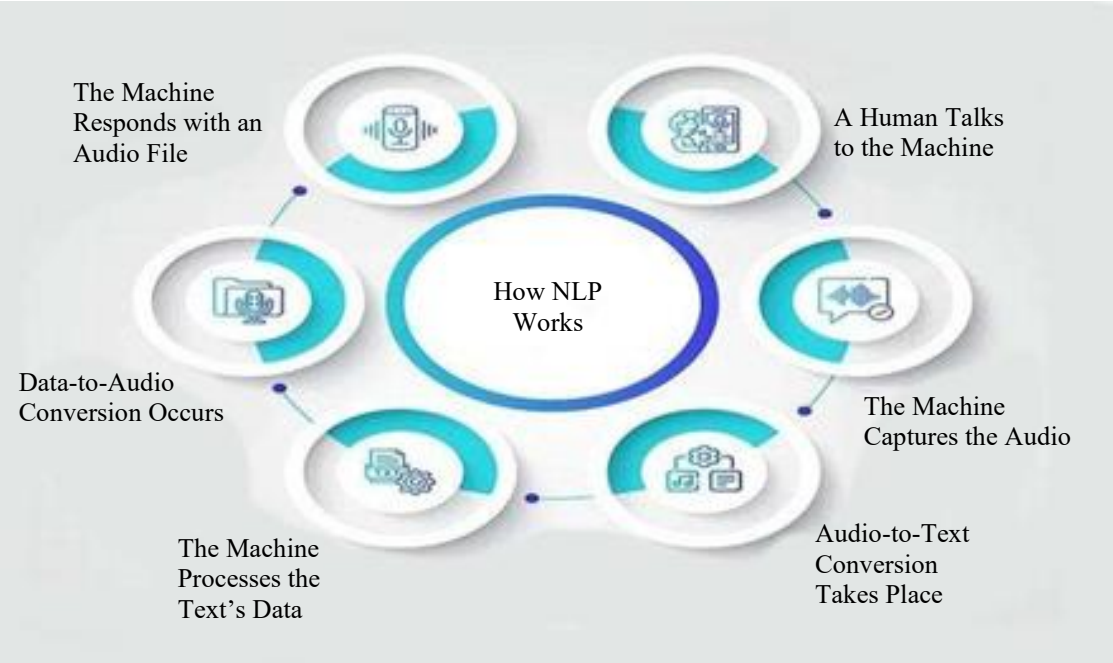

Figure 3. Workflow of Natural Language Processing (NLP) in Human–Machine Communication.

INTEGRATING PHENOTYPIC AND GENOTYPIC DATA: TRANSFORMING DIAGNOSTICS WITH AI

In the medical field, the integration of phenotypic and genotypic data has become crucial for understanding diseases, developing diagnostic tools, and personalizing treatment strategies. AI helps connect different types of medical data, like genetic information and physical symptoms, to find patterns that improve understanding and treatment of diseases. With some of the popular techniques and algorithms incorporated with AI, such as machine learning (ML), natural language processing (NLP), and computer vision, the field of medical phenotyping has seen significant advancements [18]. ML models focus on structured data patterns, NLP extracts meaningful insights from unstructured clinical records, and deep learning improves phenotyping by analyzing complex medical images and genetic data. These technologies collectively advance our understanding of genotype-phenotype relationships and improve diagnostic strategies across medical disciplines.

Role of AI in Rare Diseases

Advancements in AI technology are revolutionizing rare disease detection by analyzing patient data and assisting in accurate diagnoses. AI digs through all kinds of health and non-health data to help doctors spot and treat rare diseases. In personal healthcare, it finds genetic quirks and gives early diagnoses. Plus, it helps specialists map out treatment plans by looking at how well different drugs actually work. Overall, AI enhances decision-making for healthcare professionals. Machine learning algorithms are implemented in clinical workflows. Decision support systems enhance decision-making. AI systems aggregate feedback and improve performance. AI is used to diagnose diseases and analyze lab results [19]. Patient involvement is important. AI supports analysis bottlenecks in genetic diagnostic testing. AI models rare conditions and identifies affected pathways. Drugs are determined through in silico modeling. Analysis of image and transcriptome data is performed. Rare diseases are disregarded due to limited samples.

Types of Algorithms and Associated Benefits for Disease Diagnosis

AI algorithms have really changed the game for diagnosing rare diseases and other tough conditions [20]. Machine learning works in a few different ways.

- First, there is unsupervised learning – it just digs through data and finds patterns all by itself.
- Then there is supervised learning. This one learns from old cases and uses them to figure out new ones.

- And finally, reinforcement learning steps in. It is all about trial and error, learning what works by getting things right or wrong and picking up rewards or penalties along the way.

The earliest AI tools focused on sifting through huge datasets to help doctors make decisions when diagnosing RDs. Newer AI, on the other hand, looks for subtle clinical clues that doctors often miss early on. This shift makes a real difference – it speeds up early diagnosis, which can make prevention and intervention possible before things get complicated.

AI has become a real game-changer for diagnosing rare diseases. It spots patterns in medical images, sifts through genetic data, and gives doctors an extra layer of confidence. Machine learning is leading the charge here. We already see it catching diseases like systemic sclerosis, sometimes even beating old-school tools like high-res CT scans or lung function tests when it comes to finding lung problems. Deep learning pushes things even further. It reads images in layers, picking up on details you would never catch at first glance. With this approach, deep learning systems can handle massive, complicated datasets and spot things with a level of accuracy we just have not seen before.

AI in Rare Disease Treatments

The pharmaceutical industry has mostly ignored rare diseases because, so few people have them. For patients, that means treatment options are limited and usually expensive. Still, things are not all bleaks. Government support in recent years – backed by data from the National Institutes of Health (NIH) [21] and the US Food and Drug Administration (FDA) – has helped get more rare disease drugs approved, and AI has played a part in speeding things up. The US Orphan Drug Act (1983) and similar regulations in Europe (since 2000) have pushed companies to innovate for rare diseases. Even with these incentives, the usual drug discovery process makes progress slow and tough, since there is not much money to be made.

APPLICATIONS OF PHENOTYPIC AND GENOTYPIC MODEL

- AI digs through medical data – genetics, scans, clinical records – to spot rare diseases early on. This boosts how accurately doctors can diagnose people.
- In radiology, AI does not miss a thing. It scans X-rays and MRIs, picking up tiny details that point to rare diseases doctors might not notice at first glance.
- It finds rare genetic mutations and connects them to specific diseases.
- AI takes precision medicine seriously. It builds treatment plans tailored to each person's unique genetic profile, so patients with rare diseases get care that actually fits them.
- For drug repurposing, AI speeds up the hunt for existing medications that work against rare diseases – sometimes finding new uses for old drugs.
- Clinical trials get a boost, too. AI helps set them up, matches the right patients, and predicts outcomes, making the whole process faster and more efficient for rare diseases.
- AI-powered wearables keep tabs on people in real time. They monitor symptoms and how well treatments are working, right as things happen.
- With telemedicine, AI lets doctors remotely monitor rare disease patients. This means fewer trips to the hospital and more peace of mind for everyone [22].
- AI pulls together clinical reports, genetic data, and medical images into one big picture. This helps doctors see what is really going on and pick the best treatment.
- On the research side, AI connects isolated datasets from all over the world. This helps researchers work together, speeding up discoveries for rare disease treatments.

ECONOMIC IMPACT OF AI IN RARE DISEASES

- *Reduction in Healthcare Costs:* Bringing AI into rare disease care actually cuts healthcare costs in a big way. When doctors use AI for diagnostics and catch diseases early, patients skip a lot of those pricey, drawn-out treatments and hospital stays. Early and accurate diagnosis stops the cycle of

misdiagnoses and delays that usually rack up costs and push patients into more serious, expensive care down the road [23].

- *Lower Drug Development Costs:* AI's ability to streamline drug discovery and clinical trials can significantly reduce the cost of developing treatments for rare diseases. Traditional drug development is an expensive and lengthy process, but AI's predictive capabilities can shorten development times, cut research costs, and improve the success rates of clinical trials. These cost savings will make it more economically feasible to develop orphan drugs, which are often seen as financially unviable due to the small patient populations [24].
- *Increased Access to Treatments:* AI can reduce inequalities in access to rare disease treatments. By making diagnostics and expert consultation more widely available, AI can democratize healthcare access, particularly for patients in underserved regions or those with limited access to specialized care [25]. This could lead to a more equitable distribution of healthcare resources, ultimately reducing the overall economic burden of rare diseases on societies.
- *New Market Opportunities:* AI keeps pushing forward, and with it, healthcare is changing fast. We are starting to see fresh business models pop up – things like AI-driven healthcare platforms, smarter diagnostic tools, and personalized medicine. All of this creates new ways for companies in biotech, pharma, and healthcare to make money, and honestly, it is just the beginning [26]. Moreover, as AI enhances the effectiveness of treatments for rare diseases, it will drive investment in the rare disease market, which has traditionally been underfunded.
- *Cost of AI Implementation:* AI can save money down the road, but getting started is not cheap. Hospitals and clinics must pour a lot into AI infrastructure, data systems, and make sure their staff know how to use everything [27]. Over time, these investments usually pay off. Still, that first big expenditure can be tough, especially for places that do not have much to work with.

CHALLENGES FACED BY AI IN HEALTHCARE

AI faces several challenges in rare disease healthcare, primarily due to the limited availability of data, as these diseases affect small populations, making it difficult to train robust models. Data fragmentation across institutions, a lack of comprehensive patient registries, and complex disease mechanisms further complicate AI development [28]. Additionally, small, imbalanced datasets can lead to overfitting and reduced accuracy, while issues of trust and interpretability hinder adoption by healthcare professionals. The absence of clear regulatory guidelines and ethical concerns, such as patient consent and data privacy, also pose significant barriers to AI integration in rare disease care. Here are some key challenges faced by AI in healthcare:

- *Data Privacy and Security:* AI systems require access to sensitive patient data, raising concerns about data security, patient privacy, and compliance with regulations.
- *High Cost and Resource Requirements:* AI in healthcare does not come cheap. You need serious money, solid infrastructure, and a lot of computing power to get these systems up and running. For smaller hospitals or clinics – especially those working with tight budgets – these expenses can feel impossible to manage.
- *Shortage of Skilled Professionals:* There is just not enough people who get both healthcare and AI. Data scientists know how to build models, but most do not speak the language of medicine. On the flip side, doctors and nurses usually have not trained in AI. So, this gap slows everything down when it comes to actually putting AI to work in hospitals or clinics.
- *Regulatory and Legal Uncertainty:* Bringing AI into medical decisions opens a whole can of worms legally and ethically. Right now, nobody is really sure who is responsible if something goes wrong with an AI system – there are not clear rules yet. That leaves both healthcare workers and developers in a tough spot, unsure where the lines are.
- *Patient Trust and Acceptance:* A lot of patients do not trust AI to make calls about their health. If they do not really understand how technology works, it is easy to worry about accuracy or transparency. People want to know who is making decisions about their care, and if it is an algorithm, that can be a hard sell.
- *Scalability Challenges:* Sure, AI can look great in small pilot projects. But rolling it out on a bigger scale is a whole other story. Every hospital is different – different tech, different patients,

different routines. Making AI work in the real world takes way more resources and careful planning.

- *Ongoing Monitoring and Maintenance:* You cannot just set up an AI system and forget about it. These things need constant attention – monitoring, updates, retraining – as new medical information comes in and healthcare keeps changing.
- *Limited Data for Rare Diseases:* When it comes to rare diseases, there just is not much data out there. Not many patients, not many records or genetic details to go on. That makes it really tough to build AI tools that are actually accurate for these conditions.
- *Fragmented Data Sources:* What little data does exist for rare diseases is scattered all over the place – different hospitals, labs, and databases. Pulling it all together is a huge challenge, and that makes it even harder to create strong, reliable AI models.
- *Lack of Patient Registries:* Many rare diseases lack comprehensive patient registries, which are crucial for collecting data that could be used to train AI models and improve diagnosis and treatment outcomes.

FUTURE DIRECTIONS AND INNOVATIONS

AI is going to pull out the key details from a patient's digital records. At first, that just means things get faster and smoother for doctors. But once these systems get enough real-world testing, they will start shaping decisions about patient care directly. AI tools will not just speed things up – they will actually bring specialist-level skills right into your local clinic. Let us say a GP snaps a photo of a weird-looking mole. Instead of waiting weeks for a specialist, that image goes straight to an AI trained on dermatology and comes back with a diagnosis almost instantly. And it is not just skin stuff. AI already shows real promise reading all sorts of medical images – retinal scans, X-rays, ultrasounds. The cool part? Most of these images do not need fancy or expensive gear. Even basic clinics can handle it. Looking ahead, AI research needs to zero in on the kinds of tasks that actually match what is happening in medicine right now. But here is the thing: if we really want AI to work in the clinic, we need doctors and AI to actually work together. AI can make things cheaper and more efficient, but it still needs doctors to teach it how to handle tricky, real-life cases. That partnership matters. Most importantly, we cannot let AI take humanity out of medicine. If people start feeling like machines are running the show, they will push back. Honestly, the biggest roadblock is not the tech – it is whether people trust it enough to welcome it into their care [29].

AI in Healthcare Resource Optimization: AI-driven systems will continue to improve the management and allocation of healthcare resources, ensuring that the limited resources available in treating rare diseases are used more efficiently. By providing insights into the most effective treatments, predicting patient needs, and optimizing hospital workflows, AI will help reduce inefficiencies and lower the overall cost burden on healthcare systems, making care more accessible.

CONCLUSION

AI is not just about automating tasks. It can actually help diagnose diseases, create personalized treatment plans, and back up doctors when they are making tough calls. The real promise here is using technology to improve patient care everywhere – from big hospitals to local clinics [30]. What really sets AI apart is how quickly it can sort through huge amounts of data. That speed and accuracy open up new ways to boost patient outcomes, cut down on healthcare costs, and lift the overall quality of care. Tools like machine learning and natural language processing help doctors spot patterns they might have missed and make smarter decisions. But it is not all smooth sailing. Bringing AI into healthcare comes with some big challenges. There is the need to keep patient data private and secure, deal with any bias hiding in the algorithms, make sure AI decisions stay transparent, and figure out the regulatory maze. On top of that, people need to think seriously about ethics – like how AI will affect healthcare jobs and what it means for the relationship between patients and these new systems. Moving forward, it is going to take a team effort. Technologists, healthcare pros, policymakers, and patients all need to work together to get the most out of AI while keeping risks checked. With ongoing research, smart regulations, and everyone bringing their expertise to the table, AI can truly reshape healthcare. The future could mean care that is faster, more personal, and fairer for everyone.

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