Rare Expertise

WHITE PAPER

AI and the Future of Rare Disease Care

How Artificial Intelligence Is Reshaping Diagnosis, Drug Development, and the Clinical Workforce

A Rare Expertise White Paper

For rare disease marketing and medical affairs professionals

rareexpertise.com

Contents

SECTION 01

Executive Summary

Artificial intelligence is poised to reshape healthcare unevenly — and nowhere will that unevenness matter more than in rare disease. Rare disease diagnosis has long been described as a "diagnostic odyssey": patients often wait years, seeing multiple specialists, before receiving an accurate diagnosis. That delay is not primarily a knowledge problem. It is a pattern-recognition problem, and pattern recognition is precisely where AI excels.

This paper outlines three plausible scenarios for AI adoption in healthcare — slow, moderate, and rapid — and examines what each would mean for general health outcomes, rare disease diagnosis and treatment, and the clinical workforce. It then looks more closely at which categories of rare diseases and rare cancers stand to benefit earliest, and why.

3

AI adoption scenarios modeled

SLOW · MODERATE · RAPID

1–2yrs

projected diagnosis time in the moderate scenario

DOWN FROM 5–7 YEARS

5–10x

potential acceleration in drug repurposing

RAPID ADOPTION SCENARIO

The central thesis is straightforward: AI's nearest-term and most durable contribution to rare disease will not be replacing clinicians or delivering autonomous diagnoses. It will be compressing the time between a patient's first symptoms and a clinician's first moment of suspicion. Everything else — faster trials, expanded therapeutic options, more efficient drug development — follows from that earlier point of recognition.

SECTION 02

The Diagnostic Odyssey Problem

For most rare disease patients, the barrier to timely care is not a lack of available treatment — it is the length of time it takes to arrive at a diagnosis in the first place. Diagnostic delay for rare conditions is commonly measured in years, not weeks, and the reasons are consistent across disease areas:

- Symptoms present in fragments across multiple specialists who never see the full picture.
- Anchoring bias and premature closure lead clinicians to settle on a common explanation for an uncommon disease.
- Primary care workflows are not structured to prompt genetic or specialist workup for unusual symptom clusters.
- Data remains siloed across electronic health record (EHR) systems, limiting the aggregation of subtle, multi-system signals.

> KEY INSIGHT
>
> Rare disease diagnosis is, at its core, a pattern-recognition and data-aggregation challenge — exactly the kind of task where machine learning systems already demonstrate strong performance in narrower domains like imaging and genomics.

SECTION 03

Three Scenarios for AI Adoption in Healthcare

AI adoption in healthcare will not move at a single, predictable pace. It will vary by regulatory environment, reimbursement structure, data interoperability, and provider incentives. The following three scenarios illustrate a plausible range of outcomes over the next five to fifteen years.

Scenario 1: Slow and Fragmented Adoption (5–15 years)

AI remains largely administrative — documentation, coding, and scheduling — while clinical AI tools serve only as decision support. Liability concerns and regulatory conservatism slow the adoption of more autonomous systems, and data silos persist. The diagnostic odyssey remains largely unchanged; benefit concentrates in academic rare disease centers, while early identification in community settings lags. Workforce displacement is minimal, though new roles such as clinical AI oversight leads begin to emerge.

Scenario 2: Moderate but Structured Acceleration (Most Likely, 5–10 years)

Regulatory frameworks mature, AI tools integrate into EHRs as clinical "copilots," and payers begin reimbursing AI-enabled diagnostics. This is the scenario in which rare disease transformation becomes meaningful: EHR-based phenotype clustering, automated suspicion flags, and AI-assisted genetic test selection could plausibly compress time to diagnosis from five-to-seven years down to one-to-two years in developed health systems. On the workforce side, this represents role transformation rather than job loss — physicians shift from information gatherers to judgment supervisors.

Scenario 3: Rapid AI Infusion and Structural Disruption (5–8 years)

This scenario requires major breakthroughs in multimodal AI, strong regulatory harmonization, and the integration of genomics, longitudinal health data, and wearables. For rare disease, this is genuinely revolutionary: newborn genomic sequencing becomes standard, diseases are detected pre-symptomatically, and N-of-1 treatment protocols become feasible at scale. This scenario also carries the most significant workforce disruption, with an estimated 50–70% of current image reading potentially automated in radiology.

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AI's most realistic and durable near-term contribution is not autonomous diagnosis. ***It is triggering suspicion.***

SECTION 03

Cross-Scenario Comparison

DOMAIN	SLOW	MODERATE	RAPID
Diagnostic speed	Slightly improved	2–3x faster	Near real-time
Rare disease detection	Specialty-driven	EHR-driven flags	Genomics + AI predictive
Clinical jobs	Augmented	Reshaped	Selective displacement
Hospital utilization	Stable	Gradual decline	Significant decline
Health equity	Risk of widening	Depends on policy	Major policy battleground

Across all three scenarios, one pattern holds: administrative AI adoption is likely to become near-universal regardless of pace, diagnostic copilots are likely to become mainstream, and fully autonomous medicine remains limited even under aggressive assumptions.

SECTION 04

Which Rare Diseases Are Most AI-Ready?

AI's impact on any given rare disease depends on three variables: how rich the available data is (genomic, imaging, longitudinal EHR), how detectable the underlying pattern is, and how biologically tractable the disease is. Diseases that are genotype-driven, image-detectable, biomarker-trackable, and mechanistically well-defined will benefit earliest.

Monogenic Diseases with Recognizable Phenotypic Clusters

Clear genetic cause, multi-system EHR patterns, and frequent misdiagnosis make this category a prime candidate for diagnostic-odyssey compression.

- Fabry, Pompe, Gaucher, and Niemann-Pick disease
- MPS disorders (MPS I, II, III); hereditary angioedema; Wilson disease
- Alpha-1 antitrypsin deficiency; tuberous sclerosis complex; neurofibromatosis (NF1, NF2)

Rare Neurologic and Neurodevelopmental Disorders

Subtle early signs, characteristic MRI patterns, and distinctive seizure phenotypes make this group well suited to AI-assisted detection — including Rett syndrome, Dravet syndrome, CDKL5 deficiency disorder, and rare epileptic encephalopathies.

Rare Hematologic, Immunologic, and Cardiometabolic Disorders

Recurrent unexplained lab abnormalities and mature cardiac-imaging AI make disorders such as atypical HUS, primary immunodeficiencies, hereditary transthyretin amyloidosis, and genetic cardiomyopathies strong near-term candidates.

Even in a fully AI-enabled world, clinicians will still need to recognize ***when something doesn't fit.***

SECTION 05

AI's Role in Accelerating Rare Disease Drug Development

Beyond diagnosis, AI is beginning to influence how rare disease therapeutics are designed, tested, and brought to approval. The diseases best positioned for this kind of acceleration share four features: a known gene target, a clear molecular mechanism, a quantifiable biomarker, and amenability to modalities like gene therapy, antisense oligonucleotides (ASOs), or enzyme replacement.

- Monogenic enzyme deficiencies — better vector design, capsid optimization, and dose modeling
- Genetic epilepsies (SCN1A, SCN2A, KCNQ2, CDKL5) — variant-function prediction and ASO design
- Rare complement and immune pathway disorders — pathway modeling and combination-therapy design
- Neurodegenerative diseases with biomarkers — digital endpoint validation and trial enrichment

> HIGH-LEVERAGE OPPORTUNITY
>
> For ultra-rare, single-family mutations, AI can support rapid sequence design and toxicity modeling — moving custom, N-of-1 treatment from a theoretical possibility toward a scalable platform. This may be the single most transformative application of AI in rare disease therapeutics.

SECTION 06

Rare Cancers: An Outsized Opportunity

Rare cancers are uniquely well suited to AI because they are molecularly defined, diagnosed largely through image-based histopathology, characterized through genomic profiling, and constrained by small patient populations.

Diagnosis

AI may meaningfully improve histopathology classification for sarcomas, rare lymphomas, and rare CNS and pediatric tumors — classifying subtype, identifying molecular correlates, and predicting mutations directly from histology images.

Trial Matching

AI-driven molecular trial matching and cross-institution rare cohort aggregation could substantially improve enrollment in trials that would otherwise struggle to reach a viable sample size — possibly the largest single impact of AI in rare oncology.

SECTION 07

Three Tiers of AI Impact on Rare Disease

AI's impact on rare disease can be organized into three tiers, distinguished by how soon they are likely to materialize.

1
Diagnostic Compression
MOST IMMEDIATE

2
Trial Efficiency
MEDIUM-TERM

3
Therapeutic Expansion
LONGER-TERM, TRANSFORMATIONAL

SECTION 08

What AI Will Not Do

It is worth being direct about the limits of this technology. AI will not compress a five-year neurodegenerative outcome into six months, eliminate necessary safety observation periods, or remove fundamental biological uncertainty. Rare disease research remains constrained by the underlying biology, not merely by the speed of data analysis.

What AI can plausibly do is narrower, but still significant: reduce the rate of development failure, expand the number of rare diseases considered clinically "addressable," and — with appropriate regulatory alignment — reduce the patient numbers required to support approval.

SECTION 09

Implications for the Healthcare Workforce

Across all three adoption scenarios, the workforce story is one of transformation rather than wholesale replacement. Physicians increasingly move from being the primary generators of differential diagnoses to being the interpreters and communicators of AI-generated possibilities. Radiologists and pathologists see AI pre-read the bulk of routine images and slides, shifting their own time toward interventional work and complex case synthesis.

New roles are likely to emerge across every scenario: clinical AI trainers, data phenotype curators, model validation specialists, and algorithm safety and bias officers — professions that barely exist today but may become standard fixtures in health systems within a decade.

SECTION 10

Conclusion: Preparing for an AI-Enabled Future

The most likely path forward is the moderate, structured acceleration scenario: administrative AI adoption becomes near-universal, diagnostic copilots become mainstream, and rare disease detection improves dramatically in the health systems that adopt these tools well. The workforce is transformed, not decimated.

For rare disease specifically, the throughline across every scenario is the same: AI's most realistic and durable near-term contribution is not autonomous diagnosis. It is triggering suspicion — surfacing the pattern that prompts a clinician to pause, reconsider, and order the test that finally ends a diagnostic odyssey.

Rare Expertise

Rare Expertise is a strategic consultancy focused on helping companies developing and marketing products for patients with rare diseases. Our mission is to shorten the diagnostic and treatment journey in people with rare diseases through better education.

Rare Expertise and the Rare Medical Network work at the intersection of rare disease knowledge, clinical practice, and trusted professional networks. Our focus is on supporting healthcare professionals with credible information and access to expertise — when it matters most.

FOUNDED

Rare Expertise was founded in 2015 by Jack Davis and Jeff Sweeney, who are parents of children with rare diseases, and who both have extensive professional experience in marketing communications and medical education in rare disease markets.