Branding Science Group

Unlocking the Future of Rare Disease Care:
The Promise of AI

Artificial intelligence in the healthcare space is not a new concept, but its power can have a new meaning for rare disease.

Artificial intelligence in the healthcare space is not a new concept. As early as the 1970s, pioneering AI systems were developed to support medical diagnosis. However, these early models were limited in scope and capability, preventing widespread adoption in clinical practice. Fast-forward to today, AI is a powerful force—analysing data, learning on its own, and delivering insights faster and at greater scale than ever before..

Yet, as AI's capabilities grow, so too do concerns among healthcare professionals. Many clinicians' express unease about the "black box" nature of AI decision-making, fearing a loss of clinical autonomy and the erosion of the doctor-patient relationship. Ethical questions around data privacy, algorithmic bias, and accountability in case of error remain unresolved, prompting calls for robust regulatory frameworks.

This transformation is especially impactful in rare diseases—a field long hindered by diagnostic delays, scarce treatments, and fragmented care. For the 300 million people affected worldwide, diagnosis often takes years, marked by misdiagnoses and emotional strain. The clinical and economic burden is substantial, with repeated consultations and delayed interventions.

Recent breakthroughs in AI, especially in deep learning and natural language processing, are beginning to change this narrative. Tools like BioClinicalBERT (and even general-purpose models like ChatGPT) have demonstrated the ability to extract critical disease characteristics from unstructured medical records with minimal training, accelerating the diagnostic process. At the same time, AI is proving invaluable in analysing vast datasets, from genomic sequences to wearable device outputs, enabling more tailored and precise treatment strategies.
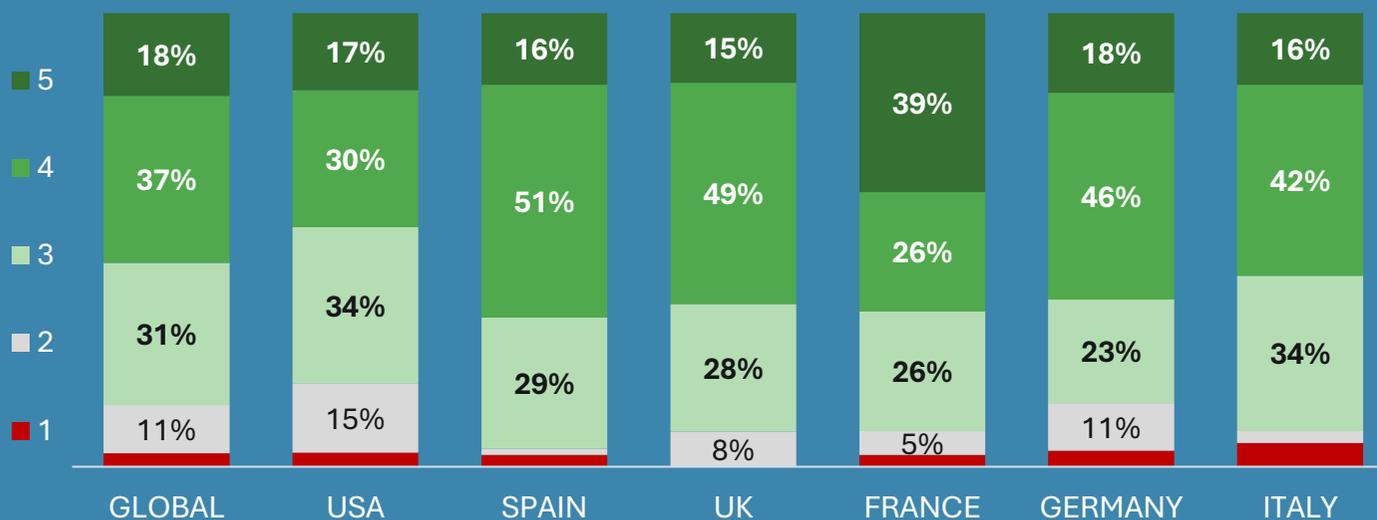
By harnessing AI's capacity to detect patterns invisible to the human eye, healthcare providers can not only improve diagnostic accuracy but also anticipate therapeutic responses and even implement preventative measures. In rare disease care, where every moment counts, AI may well be the solution patients and clinicians have long awaited.

AI Sparks Hope: Physicians treating rare diseases show strong enthusiasm for AI's role in future care
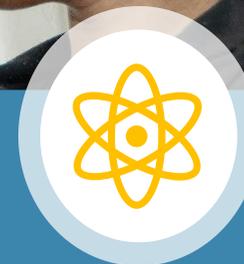
Using a rating scale where 1 = I do not expect to use any AI tools at all, and 5 = I already extensively use AI tools, how open would you be to using AI tools to support you in your work managing/ treating patients?

Rating	GLOBAL	USA	SPAIN	UK	FRANCE	GERMANY	ITALY
5	18%	17%	16%	15%	39%	18%	16%
4	37%	30%	51%	49%	26%	46%	42%
3	31%	34%	29%	28%	26%	23%	34%
2	11%	15%		8%	5%	11%	
1							

Physicians specialising in rare diseases express strong optimism about the transformative potential of artificial intelligence in advancing the development and management of these conditions. An overwhelming 97% believe AI could play a meaningful role in this space, with over 85% either currently using or open to using AI in their practice, significantly higher than the 65% adoption rate among non-rare disease treaters. This enthusiasm is particularly pronounced among physicians in France, followed by those in the UK and Spain.

Across Europe, Rare Disease physicians see promise of **smarter diagnoses, better research, and improved patient outcomes with AI**

The perceived benefits of AI are most notable in areas such as pattern recognition (56%), support for clinical research (53%), and the acceleration of diagnostic processes (51%).

Additionally, 48% of physicians see AI as a valuable tool for reducing diagnostic errors by highlighting areas that warrant further investigation. Rare disease specialists are more attuned than non-specialists to the ways AI can enhance both the management and advancement of care in this complex field.

Adoption and openness towards AI vary across countries. French physicians are especially optimistic about AI's diagnostic capabilities, with 66% citing its potential to speed up diagnosis and 58% recognizing its ability to reduce diagnostic errors. Furthermore, 55% see AI to support treatment decision-making. In contrast, UK physicians are most enthusiastic about AI's role in accelerating drug development (67%) and enhancing clinical research (74%), particularly through the analysis of large datasets to identify novel therapeutic targets. Italian physicians, however, remain more cautious, especially regarding AI's utility in clinical use beyond clinical research.

How do you think AI could be able to support management / future development in rare diseases/conditions?

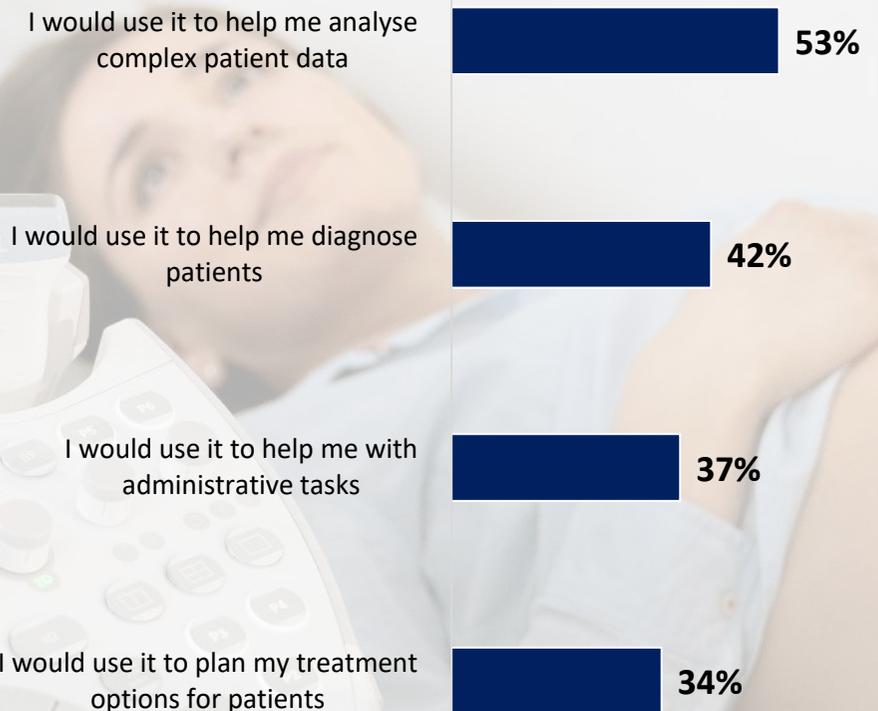|	
I think it could help with pattern recognition of complex & varied datasets (patient results, symptoms, genetic information…)	**56%**
I think AI could facilitate clinical research (analyse large datasets from clinical trials and research studies to uncover new insights into rare diseases)	**53%**
I think it could help with accelerating diagnosis	**51%**
I think it could help reducing diagnostic errors by providing a second opinion or flagging potential issues that require further investigation	**48%**
I think AI could enhance drug development (i.e identify drug candidate, analysing biological data to predict compounds in RD, speed up development)	**46%**

Physicians also see AI as a potential to **diagnose complex cases and reduce admin burden**, especially in UK and US

When asked about practical applications, physicians most frequently identified "analysing complex patient data" (53%) and "assisting in diagnosis" (42%) as the top areas where they would personally use AI. Beyond clinical functions, non-clinical benefits are also recognised: 37% of healthcare professionals see AI as a tool to alleviate administrative burdens, potentially reducing staff burnout and freeing up time for more complex, value-driven and patient-facing tasks . This view is especially prevalent among physicians in the UK and US (46% each), while only 9% of Italian physicians share this sentiment.

Thinking about where AI could support you in your practice (if the right tool was developed), please could you pick the top 3 areas where it could have the most impact for you?

I would use it to help me analyse complex patient data **53%**

I would use it to help me diagnose patients **42%**

I would use it to help me with administrative tasks **37%**

I would use it to plan my treatment options for patients **34%**

However, physicians remain cautious as AI's promise in healthcare is shadowed by concerns over errors, bias, and data gaps
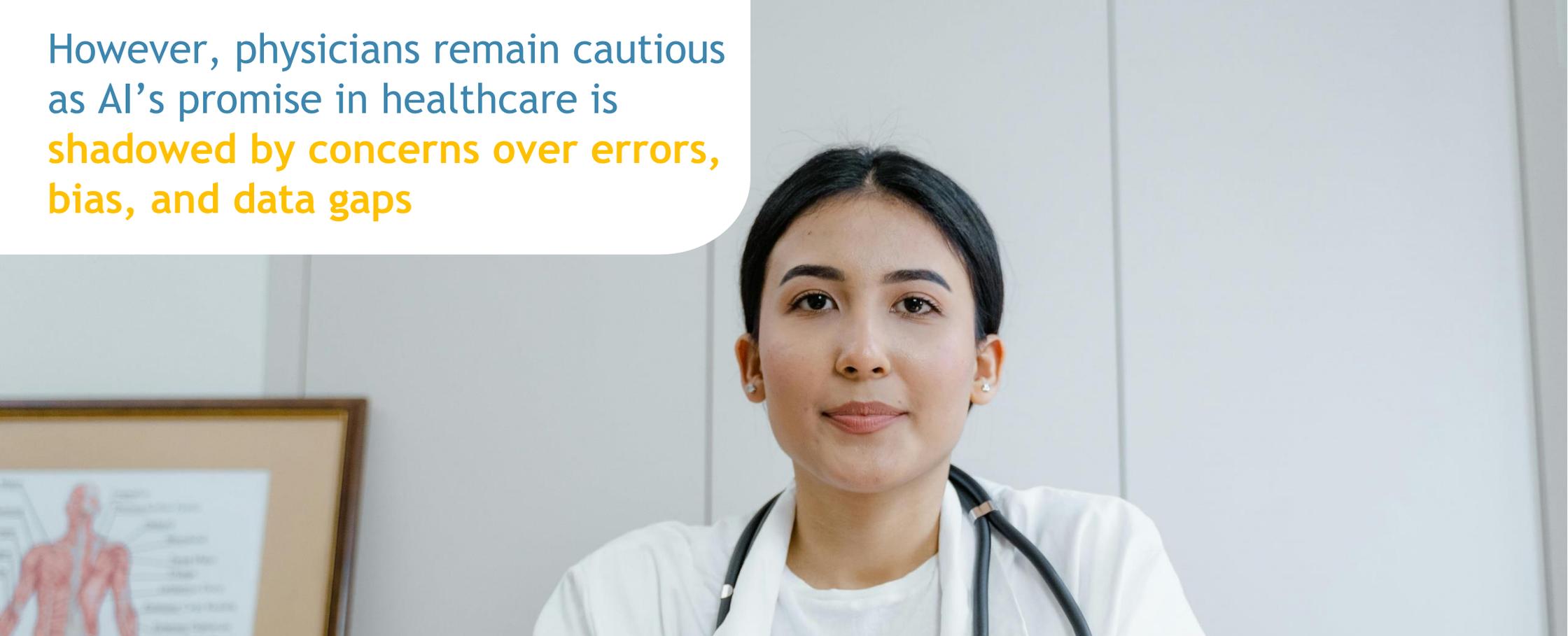
While the promise of AI in healthcare is undeniable, a degree of caution persists among physicians—particularly those outside the rare disease field and among paediatricians, with 35% and 24% respectively stating they do not intend to use AI now or in the future. This hesitancy is understandable given the high stakes: errors in AI outputs could not only harm patients but also deepen health disparities and erode trust in healthcare systems.

Concerns around AI's reliability are well documented. Physicians cite issues such as data bias, lack of robustness, and hallucinations — errors that can result in fabricated or inaccurate results with serious clinical consequences. Despite industry assurances that hallucinations will diminish over time, recent evidence suggests they remain a persistent challenge. As New Scientist aptly noted, "a model that consistently states falsehoods and requires fact-checking won't be a helpful research assistant."

Beyond hallucinations, experts like Princeton's Arvind Narayanan warn that AI models often rely on outdated or unreliable sources. In the UK, data privacy concerns are further compounded by the fact that around 6% of the population has opted out of having their health data used for research, according to MedConfidential. In the rare disease space—where data is already limited—such gaps can further hinder AI's learning and effectiveness

Base: 541 HCPs (Paediatricians, Dermatologists, Neurologists, Endocrinologists, Haematologists) in UK(42), FR(45), IT(49), ES (71), DE(47) + US (287) – all treating rare or ultra-rare diseases

Rare disease physicians cite accuracy, privacy, and liability risks amid rising cybersecurity threats as key worries

These concerns are reflected in the attitudes of rare disease specialists themselves : 46% express doubts about AI model accuracy, and 35% question the trustworthiness of the data used to train them. Privacy and security also remain top concerns across all markets, though less so in Italy.

The growing threat of cyberattacks adds another layer of risk. As The Lancet warns, it's not a matter of *if* but *when* such breaches will occur. With more healthcare systems digitising sensitive data and tools, inadequate security measures leave them vulnerable. The consequences are real: stolen or lost data, cancelled appointments, and compromised patient care. These risks are especially salient in the UK, where 74% of physicians express concern—likely influenced by recent ransomware attacks on NHS trusts.

Finally, in an increasingly litigious environment, the question of liability looms large. Could physicians be held accountable for AI-driven errors? This ethical and legal uncertainty is particularly pronounced in France, where one in three physicians report hesitancy to adopt AI due to these concerns.

What reasons could make you hesitant to use AI to support you in managing/treating patients?

Reason	%
I would need to be reassured about data privacy/security before sharing patients' data	51%
I am unsure of the accuracy of the model (what type of data was used to train the model)	47%
I am unsure I can trust the data	37%
I have ethical/legal concerns about using AI in healthcare	26%

A call for responsible progress and a strategic imperative to transform care

AI holds transformative promise for rare disease care, enabling faster diagnoses, deeper insights, and more personalised treatments. But integration into clinical practice brings challenges. Concerns around data integrity, privacy, and ethical accountability must be addressed through strong safeguards and frameworks. AI should enhance—not replace—clinical judgment.

As the British Medical Association notes, AI's success depends on thoughtful integration. Collaboration among clinicians, technologists, regulators, and patients is vital, supported by transparent governance, rigorous validation, and ongoing education.

The opportunity is clear: AI can close diagnostic gaps, ease clinician burden, and improve outcomes. But trust must be earned. Now is the time to shift from cautious optimism to responsible action — ensuring AI drives ethical, equitable, and patient-centred care.

In rare diseases, the stakes are even higher. With small patient populations and limited data, timely diagnosis and treatment are often out of reach. AI offers a unique chance to overcome these barriers, making rare disease care a priority for innovation.

Clinicians in this field are often open to new tools, recognizing AI's potential to accelerate research and improve care. But the risks of inaction are significant: without AI, delays, misdiagnoses, and missed treatment opportunities may persist, perpetuating the very inequities AI could help resolve.

Therefore, AI in rare disease should not be treated as a niche application, it must be a strategic priority. By focusing efforts where the impact is greatest, we can ensure that AI delivers on its promise not just broadly but deeply transforming care in areas where it's most needed.

GET IN TOUCH

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