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AI, Big Data and rare diseases

(exploratory opinion requested by the Danish Presidency of the Council of the EU)

(C/2026/24)

Rapporteur: **Juliane Marie NEIIENDAM**

Advisor	Marine CORNELIS (Advisor to the rapporteur, Group III)
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1. Conclusions and recommendations

1.1. The EESC welcomes how artificial-intelligence (AI) can optimise rare disease patient pathways and support the development of personalised medicines and rare disease treatment to improve the health and quality of life of rare disease patients. AI and Big Data have the potential to revolutionise rare disease treatment while safeguarding patient rights and ensuring gender-inclusive healthcare innovation. However, the application of AI and Big Data in rare disease research raises serious concerns about data privacy, algorithmic bias, affordability and geographical accessibility. To mitigate these concerns, the EESC has a range of recommendations to ensure that AI and Big Data improve rare disease diagnosis and treatment while maintaining high ethical and legal standards.

1.2. The EESC encourages all EU Member States to digitise their health data as soon as possible, as better-quality data registration standards are required for the optimal functioning of the European Health Data Space (EHDS) and advancing research. The EESC recommends the national use of ORPHA codes to ease cross-border data sharing.

1.3. In implementing the EHDS, Member States should ensure that AI healthcare models are granted access only to anonymised and encrypted patient data, thereby preventing misuse. Any misuse of health data must be subject to proportionate and dissuasive sanctions. Furthermore, the EESC recommends that Member States establish clear patient consent frameworks for AI health data usage and independent monitoring bodies to audit AI health projects and ensure transparency.

1.4. The European Medicines Agency should require explainability in augmented intelligence based healthcare decisions to ensure transparency, and physician oversight in medical recommendations and applications should be mandatory.

1.5. Professional training on AI-driven diagnostic tools should be developed to support rare disease care and ensure human oversight. The EU, in cooperation with the Member States, should develop policies to promote training schemes in collaboration with vocational bodies, businesses, research centres, social partners and civil society. EU-wide campaigns should raise AI literacy among patients and healthcare professionals to improve awareness of AI's role in rare disease diagnosis and treatment.

1.6. AI healthcare models should be trained on diverse, gender-balanced datasets to avoid discrimination. The EESC recommends that the EU AI Office should promote gender-diverse training data, bias audits and pre-market gender testing for medical AI in cooperation with the European AI Board. The EU Digital Education Action Plan should provide targeted funding for women in AI and medical data science.

1.7. The European Institute of Innovation and Technology (EIT) should expand mentorship and leadership programmes to increase female participation in AI-driven healthcare. The Horizon Europe Health Cluster should fund gender-focused AI research to develop better diagnostic tools for conditions that disproportionately affect women and underrepresented groups (e.g. autoimmune diseases, rare cancers).

1.8. Data governance structures must allow patients to control their data, including the right to withdraw consent and understand how their data is used. Consent must be ongoing, not a one-time formality, keeping power with the patient, especially before the sale of AI-processed health data to third parties. Models of data cooperatives or patient-led registries (like MIDATA in Switzerland ([1])) are gaining traction as alternatives to corporate or state data monopolies.

1.9. The EU should dedicate funding under Horizon Europe for startups and SMEs developing AI for rare disease diagnostics to prevent dominance by large corporations. Horizon Europe and the Digital Europe Programme should support affordable, accessible AI tools through public-private partnerships. Fair data access must be ensured, and AI-driven health innovations should remain publicly accessible.

1.10. The EESC welcomes the AI development of personalised medicines and rare disease treatments but highlights the need for sustainable price-setting mechanisms that respect national competences. The public investment in medicine development requires fair and sustainable pricing of rare disease therapies. The EESC recommends creating a publicly funded AI-driven diagnostic platform for rare diseases under the EU4Health Programme, and implementing cross-border AI data-sharing to allow smaller hospitals and research institutions to benefit from AI advancements.

1.11. Finally, EU-wide ethical guidelines for AI in healthcare should be established, ensuring equal access and patient safety.

2. **Introduction**

2.1. Collectively, an estimated 5 % of the population have a genetic disease. Rare diseases affect fewer than 1 in 2 000 people, yet, with more than 7 000 types of rare disease in existence, the burden worldwide is not insignificant ([2]). To date, approximately 300 million people live with rare diseases, including 30 million people in the European Union ([3]).

2.2. Around 80 % of rare diseases have a genetic cause, almost 70 % of which present in childhood, and about 30 % of children with a rare disease die before the age of five. Due to their low prevalence, patients often face significant challenges, including an average diagnostic delay of 4-5 years, limited treatment options, and a lack of coordinated care. According to Eurordis – Rare Diseases Europe, 95 % of rare diseases have no approved treatment, leaving millions without adequate medical support.

([1]) MIDATA.
([2]) The landscape for rare diseases in 2024.
([3]) Rare diseases – European Commission.

2.3. AI and Big Data offer transformative potential in addressing these challenges in terms of optimising rare disease patient pathways by improving early detection, enabling personalised treatments, and supporting innovative research. AI-driven diagnostic tools can analyse medical images and genetic data to detect rare diseases with up to 90 % accuracy in some cases ([4]). Similarly, Big Data enables the integration of patient registries across EU countries, helping researchers identify new care pathways.

2.4. The EESC has consistently expressed solidarity with the rare disease community by adopting opinions ([5]) ([6]), hosting conferences (March and October 2023, November 2024 and April 2025 ([7])), and stimulating information exchange on rare diseases. It has continuously supported the call for a European action plan on rare diseases, which should highlight the opportunities with artificial intelligence to optimise patient care pathways and research.

2.5. The EU has already taken steps to leverage new technologies while safeguarding patient rights:

— The European Health Data Space (EHDS) and the European Reference Networks ([8]) (ERNs) aim to securely share health data across Member States to advance rare disease research and treatment.

— ERNs are cross-border networks connecting European centres of expertise to improve rare disease care by enabling specialists to consult on complex cases and recommend accurate diagnoses and treatments ([9]). The Joint Action JARDIN ([10]), with EUR 18,75 million in funding until 2027, supports integration of ERNs into national healthcare systems.

— The EU AI Act establishes guidelines for high-risk AI applications in healthcare to ensure transparency and patient safety.

— The General Data Protection Regulation (GDPR) enforces strict privacy standards essential for AI-driven rare disease research.

— The EU Pharmaceutical Strategy calls for better regulatory frameworks to encourage innovation and research in rare disease treatments.

3. **Opportunities and potential of AI and Big Data in rare diseases**

3.1. AI and Big Data can significantly improve rare disease diagnostics and patient care pathways by identifying patterns in vast datasets, leading to earlier and more accurate diagnoses. Studies suggest that AI-assisted diagnostics can reduce diagnostic errors by up to 40 % ([11]), while machine learning models can shorten the average diagnostic journey by at least one year ([12]).

([4]) A look at AI-driven medtech for rare disease diagnosis.
([5]) OJ C, C/2025/115, 10.1.2025, ELI: http://data.europa.eu/eli/C/2025/115/oj, OJ C, C/2025/105, 10.1.2025, ELI: http://data.europa.eu/eli/C/2025/105/oj, OJ C 75, 28.2.2023, p. 67 and OJ C, C/2024/879, 6.2.2024, ELI: http://data.europa.eu/eli/C/2024/879/oj.
([6]) Rare disease opinions.
([7]) Rare disease conferences.
([8]) European Reference Networks.
([9]) Public health – ERNs.
([10]) Integrating European Reference Networks into national health systems.
([11]) AI in Healthcare to Reduce Diagnostic Errors by 40 % | Keev Capital.
([12]) Reducing diagnostic delays using machine learning.

3.2. Key EU initiatives such as Horizon Europe, the EHDS, the ERNs and the EU AI Act offer a strong regulatory foundation for leveraging AI in rare disease research. The European Disability Strategy 2021-2030 emphasises the role of AI in assistive technologies, which must be further integrated into national healthcare systems. Additional funding for the development of AI support systems may alleviate some of the current disability challenges and improve quality of life of rare disease patients, given that 8/10 people with rare diseases live with disability ([13]).

3.3. Different pilot projects funded by the European Commission focus on AI usage for rare diseases (such as CoMPaSS-NMD, ERAMET, Recon4IMD and Screen4Care) and the European Health and Digital Executive Agency provides grants to stabilise and further increase the opportunities that ERNs are creating for the treatment of patients, including exploration of AI in better future therapeutic and care options. These grants should be encouraged to support the pilot projects thus promoting sustainable and structural support for the Member States and institutions involved.

3.4. The diagnostic delay for rare diseases is 4 to 5 years ([14]), often with multiple misdiagnoses, and in some cases, it can take over a decade, leading to psychological and human consequences for patients and significant health costs. AI-based tools can accelerate diagnosis by analysing genetic data, medical records, and imaging scans, identifying patterns that humans might miss ([15]). This is particularly crucial for children with rare genetic diseases, allowing for early intervention, or for patients affected by a syndrome without a name (SWAN).

3.5. 95 % of rare diseases lack approved treatments ([16]). AI can speed up drug discovery by predicting how different compounds will interact with disease-causing mutations, significantly reducing the time and cost involved in developing new therapies. AI has, for example, identified new treatments for amyotrophic lateral sclerosis (ALS) that outperformed existing treatments in lab tests.

3.6. AI can tailor treatments to individual patient profiles, ensuring better efficacy and fewer side effects. Neuroprosthetics are already being tested for rare neuromuscular diseases, providing new rehabilitation possibilities. Example: AI-powered exoskeletons, such as ATLAS 2030 ([17]), are helping children with spinal muscular atrophy (SMA) regain mobility.

4. **Key challenges and risks**

4.1. Despite its promise, the application of AI and Big Data in rare disease research raises serious concerns about control by humans, data quality, data privacy, algorithmic bias, affordability and accessibility. An overreliance on AI may lead us to neglect a holistic approach to treatment strategies, and ethical considerations. AI may lead to rational decisions that are inappropriate for the patient's situation as not all parameters are included in the AI models. The final decision must remain with the healthcare and medical profession.

4.2. Ensuring equal access to AI-driven healthcare innovations:

4.2.1. The EU Pharmaceutical Strategy for Europe emphasises the need to foster innovation in rare disease treatments and AI-driven drug discovery, especially for rare diseases that lack commercial incentives for research. These treatments are excessively priced and weigh heavily on health budgets.

4.2.2. Rare disease data in Europe is scarce and fragmented, limiting large-scale AI analysis. Without standardised high-quality data, AI may only benefit well-represented diseases or wealthy populations. Access to AI tools is uneven, with rural-urban divides, digital health infrastructure gaps between countries and high treatment costs deepening inequalities. Ensuring access to quality data is essential for effective AI-driven treatments.

([13]) Recognising the disabilities of those living with rare diseases.
([14]) Survey reveals lengthy diagnostic delays for rare disease patients.
([15]) Efficiency of computer-aided facial phenotyping (DeepGestalt).
([16]) Expanding research into rare diseases.
([17]) Marsi Bionics | Enabling human walking.

4.3. Bias and gender disparities in AI models:

4.3.1. Women are disproportionately affected by autoimmune and chronic rare diseases and wait an average of four years longer to receive a diagnosis for the same disease as men ([18]). Most preclinical studies use data from European-descent males ([19]), leading AI models trained on these datasets to misdiagnose women more frequently ([20]). An unfortunate trend in abusing women's health data is also seen ([21]). AI-based health monitoring apps often track and store sensitive reproductive health data, raising privacy concerns ([22]).

4.3.2. Most adult drug treatments offer a uniform dosage for both men and women, despite known differences in body weight, composition and metabolism, which risks overmedicating women. Women often experience higher blood concentrations of drugs and longer elimination times, leading to approximately double the rate of adverse reactions compared to men ([23]). AI models may accentuate this problem but can also help provide a differentiated drug regime.

4.3.3. AI tools trained on limited or skewed datasets may fail to diagnose rare diseases in underrepresented ethnic or socio-economic groups. They may miss atypical symptoms in women, children, or racialised communities, and they may reproduce systemic biases from healthcare systems and clinical encounters, for example a lack of belief in some patient categories, affecting who gets diagnosed and referred for treatment. A study shows black women are 40 % less likely to be diagnosed, and are diagnosed on average 2,6 years later than white women ([24]). The EU Gender Equality Strategy 2020-2025 highlights the need for gender-inclusive healthcare technologies and calls for gender-sensitive AI research to avoid discrimination in healthcare.

4.4. Data privacy and security risks:

4.4.1. AI models require vast amounts of patient data to function effectively, raising concerns about data privacy, ownership, informed consent, and security breaches. Rare disease data is highly fragmented across Europe, making it difficult to conduct large-scale AI analyses. The EHDS and the ERNs aim to facilitate secure and standardised data-sharing, but data quality, technical interoperability and patient consent remain challenges. The introduction of ORPHA codes helps, but they are not used consistently across Europe ([25]).

4.4.2. Current consent frameworks are often ill-suited for long-term AI use, especially when data is reused beyond its original purpose. Patients are rarely informed about future applications at the time of data collection. This is particularly sensitive in rare disease communities, where patients may feel a strong sense of solidarity or urgency to share their data. Without safeguards, this risks data colonialism, i.e. extracting sensitive data without fair benefits in return.

4.4.3. GDPR protects personal health data, but AI-driven research often requires cross-border data pooling, necessitating clear ethical guidelines on data ownership and privacy. To truly serve rare disease communities, AI must be governed with transparency, accountability, and respect for patients' rights. The mention of the use of AI should be clearly displayed and data must be encrypted.

4.5. Governance: The use of AI and Big Data in rare diseases is at a critical juncture. While these technologies offer faster diagnoses and better treatments, they risk reinforcing unequal power dynamics between patients, healthcare systems, and private actors. Without clear governance, rare disease data, often from vulnerable patients, could become a commodity controlled by a few tech firms or institutions.

([18]) Gender Data Health Gap Report.
([19]) Rare disease day 2025 reflecting on a year of progress and the challenges ahead.
([20]) Are AI tools failing women's health?.
([21]) Female health apps misuse highly sensitive data.
([22]) Pregnancy tracking tech with *privacy not included warning.
([23]) Sex Differences in Pharmacokinetics.
([24]) Time of diagnosis across racial and ethnic groups.
([25]) ORPHA codes.

4.6. The EU AI Act classifies healthcare AI as high-risk, requiring models to be explainable and auditable. Similarly, the GDPR protects health data. Nevertheless, some key questions arise: Who owns the data? Who decides how it's used or monetised? And who benefits? Without strong public oversight, in line with the existing EU legislative framework, patients may lose control over their most personal information to AI systems they can't understand or challenge.

4.7. AI can be a powerful tool in narrowing down possible diagnoses, especially for rare or complex diseases, but medical decisions must always include human oversight. Healthcare professionals should critically assess AI-generated recommendations before clinical action is taken. Past errors, such as unsafe cancer treatment suggestions by IBM's AI Watson for Oncology ([26]), highlight the need for reliability and accountability in AI-driven healthcare. Promoting AI transparency and helping both healthcare professionals and patients understand how AI generates diagnostic output is key to building trust and informed decision-making.

4.8. Economic barriers & SME challenges:

4.8.1. Developing AI-driven medical technologies and models for rare diseases requires high computational power and access to vast datasets, which is expensive and creates barriers for small and medium-sized enterprises (SMEs). Training a state-of-the-art AI model can cost over EUR 10 million ([27]), making it inaccessible for SMEs.

4.8.2. Large tech companies dominate AI-driven healthcare research, limiting competition. The Innovative Health Initiative (IHI) supports public-private collaboration, but further incentives are needed to encourage AI-driven medical research. The EHDS should be the accelerator of shared safe data exchange across Europe and between companies.

4.8.3. The EU can support SMEs and startups using translational research in developing AI for rare diseases through targeted funding, such as Horizon Europe. While its Health Cluster supports medical AI, it lacks a rare disease focus.

4.8.4. The EU's Digital Innovation Hubs aim to support SMEs, but dedicated AI funding is limited and insufficient. The Digital Europe Work Programme 2025-2027 has allocated EUR 90 million to AI in healthcare, but the global gene therapy market is expected to be worth USD 19,88 billion by 2027 ([28]). The Horizon Europe Health Cluster supports AI innovations in rare disease diagnosis, but funding must be increased for clinical deployment.

5. **Impact of AI on healthcare workers and employment**

5.1. The use of AI and Big Data in rare diseases will reshape the roles of healthcare professionals. Where AI will automate certain tasks, it will also augment healthcare workers' roles, enabling them to focus on more complex and human-centred aspects of care. To ensure that healthcare professionals can effectively use AI-driven tools while maintaining human oversight, tailored training and up- and reskilling programmes should be developed. These should cover not only technical skills but also ethical and safety considerations and should be designed in collaboration with vocational training bodies, businesses, research centres, social partners, and relevant civil society organisations.

5.2. Women remain underrepresented in AI and medical data science, comprising only 22 % of AI professionals globally ([29]) and remain underrepresented in medical AI research. This gender gap may lead to biased algorithms, limited female perspectives in research, and healthcare inequalities.

([26]) Watson Supercomputer Recommended Unsafe Treatments.
([27]) The Cost of Implementing AI in Healthcare: Tips and Insights for 2025.
([28]) 8 things you should know about gene therapy.
([29]) Act now to close the digital gender gap in AI.

5.3. Trade unions and worker representatives should be involved in AI governance through social dialogue to ensure workers' rights and meaningful participation in line with the features of national industrial relations systems. Additionally, the benefits and risks to health and safety arising from AI systems need careful assessment, and any risks should be appropriately mitigated. These factors are mentioned in the EESC opinion on *Health and safety at work – current and future challenges in light of traditional and new technologies, with a focus on AI* ([30]), point 2.5 and 2.6.

5.4. The EESC has previously addressed AI in the labour market in different opinions ([31]) ([32]) ([33]). The EESC continues to monitor the impact of AI on the labour market and make recommendations on how to constantly update skillsets and ensure a fair and just collaboration between employers, employees and civil society with regard to the increasing role of AI in the EU.

Brussels, 18 September 2025.

The President
of the European Economic and Social Committee
Oliver RÖPKE

([30]) Opinion of the European Economic and Social Committee – Health and safety at work – current and future challenges in light of traditional and new technologies, with a focus on AI (exploratory opinion requested by the Polish Presidency of the Council of the EU) (OJ C, C/2025/2958, 16.6.2025, ELI: http://data.europa.eu/eli/C/2025/2958/oj).

([31]) Opinion of the European Economic and Social Committee – Pro-worker AI: levers for harnessing the potential and mitigating the risks of AI in connection with employment and labour market policies (own-initiative opinion) (OJ C, C/2025/1185, 21.3.2025, ELI: http://data.europa.eu/eli/C/2025/1185/oj).

([32]) Opinion of the European Economic and Social Committee – Fostering opportunities and managing risks from new technologies for public services, the organisation of work and more equal and inclusive societies (exploratory opinion requested by the European Commission) (OJ C, C/2025/114, 10.1.2025, ELI: http://data.europa.eu/eli/C/2025/114/oj).

([33]) Opinion of the European Economic and Social Committee – Working time, the efficiency of the economy and the well-being of workers (including in the context of digital change and work automation): a legal and comparative analysis of the situation in EU Member States (exploratory opinion at the request of the Polish presidency) (OJ C, C/2025/2960, 16.6.2025, ELI: http://data.europa.eu/eli/C/2025/2960/oj).