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Protected: Ways to apply digital and technological solutions to benefit people with rare diseases



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Michael Damiano

Head of Rare Diseases UK and Ireland, Sanofi

Silvia Cerolini

Head of Innovation in Specialty Care UK and Ireland, Sanofi



Pursuing solutions and not settling for the status quo is the manuscript being written, and wielding the pen is a pharmaceutical company inspired by innovation.

Sanofi's ambition to become the first artificial intelligence (AI) driven global pharmaceutical company marks the next chapter in its commitment to healthcare professionals and patients.



AI and digital solutions in rare diseases

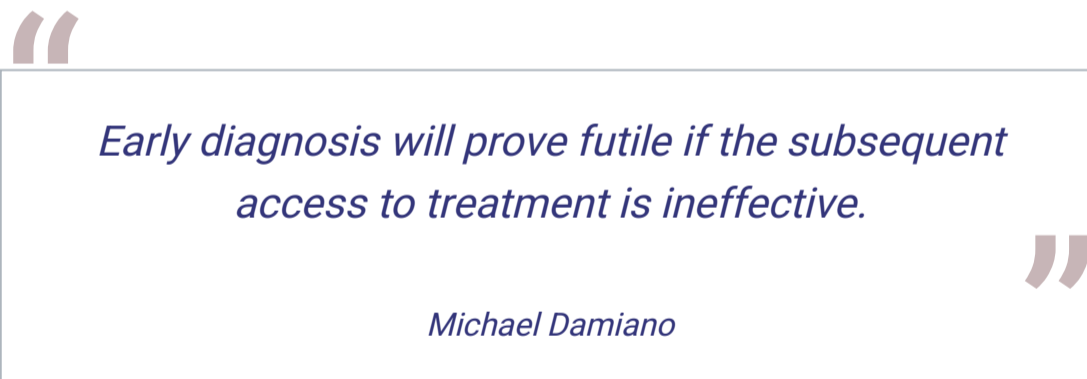
With an aim to ensure technological ambition is matched by tangible output, Silvia Cerolini, Head of Innovation in Specialty Care UK and Ireland at [Sanofi UK](#), explains: “In the UK, we’re building a blueprint to enable a life sciences ecosystem that best supports those impacted by a rare disease. By leveraging ever-evolving technologies, we can overcome hurdles currently presenting in the diagnostic journey and unlock benefits for all patients.”



Accelerating diagnosis with data and AI

As it stands, there are no commonplace methods for measuring the diagnostic odyssey, defined as ‘the time between symptoms first presenting and people receiving a definitive diagnosis.’¹ However, research has found that more than a third of people living with a rare condition will wait over five years to receive a diagnosis.²

Cerolini raises the importance of supporting healthcare professionals (HCPs) by lightening their diagnostic burden through proactive programmes. Cerolini welcomes [collaborative efforts to accelerate diagnosis](#) for patients with rare diseases by using data as a conduit to help identify those at risk. “By using expert input, this technology can be refined to help more people receive a timely diagnosis which, in turn, can prompt early intervention and better patient outcomes,” she explains.



Collaboration and partnerships

“Working alongside like-minded organisations is important for enacting a culture of curiosity,” Michael Damiano, Head of Rare Diseases UK and Ireland at Sanofi, outlines. Mendelian is a pioneer within the AI sector, recently being awarded an NHS ‘AI in Health and Care Award.’³

Their platform, MendelScan, uses AI to identify patterns of symptoms that could suggest a rare disease. “By learning from the specific expertise of different stakeholder groups, innovation across the industry can flourish. To put it succinctly, a rising tide lifts all boats,” Damiano shares.

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Enhancing care

The [intersection between life sciences and innovation](#) is increasingly recognised as a catalyst for improving the patient experience. Further progress, Damiano identifies, “requires observing the patient journey with a holistic mindset, as efforts to improve early diagnosis will prove futile if the subsequent access to treatment is ineffective.”

In fact, for every 100 patients that get a new medicine in its first year of launch in EU countries, just 21 patients in the UK will.⁴ Helping to develop a more harmonious pathway for patients requires the collaboration of industry partners and patients, with a focus on monitoring and data collection.



Commitment to the rare disease journey

“Our journey is steeped in steadfast commitment, helping to facilitate early diagnosis and improved access to treatment. This legacy is an accomplishment we are deeply proud to hold,” explains Damiano. “But, as with most great legacies, it continues to be written.”

[Click here to learn more](#)

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[1] Department of Health & Social Care. England Rare Diseases Action Plan 2023: main report; February 2023. Available at: <https://www.gov.uk/government/publications/england-rare-diseases-action-plan-2023/england-rare-diseases-action-plan-2023-main-report>.

[2] Genetic Alliance. Good Diagnosis: Improving the experiences of diagnosis for people with rare conditions; February 2022. Available at: <https://geneticalliance.org.uk/wp-content/uploads/2022/02/Rare-Disease-UK-Good-Diagnosis-Report-2022-Final.pdf>.

[3] NHS England. AI in Health and Care Award winners; March 2023. Available at: <https://transform.england.nhs.uk/ai-lab/ai-lab-programmes/ai-health-and-care-award/ai-health-and-care-award-winners/>.

[4] Association of British Pharmaceutical Industry. Improving Access to Medicines in the UK; November 2021. Available at: <https://www.abpi.org.uk/value-and-access/uk-medicine-pricing/improving-access-to-medicines-in-the-uk/>.



Author
Michael Damiano & Silvia Cerolini

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