



# **THE CASE FOR A NATIONAL CLINICAL DIRECTOR FOR RARE DISEASES**

Advancing Clinical Leadership  
to Improve Outcomes for Patients

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# EXECUTIVE SUMMARY

The UK is home to 3.5 million people living with a rare disease, and there is an opportunity to give these conditions better representation at the highest levels of NHS leadership. To accelerate research, ensure equitable access to care and improve patient outcomes, the NHS should establish a **National Clinical Director (NCD) for Rare Diseases**. This role would provide strategic leadership, raise the profile of rare diseases, and align policy and service planning with the needs of patients and healthcare professionals (HCPs). In turn, this would foster valuable opportunities for life sciences investment in research and development, further enabling the UK to harness the unprecedented opportunity to align with Government plans for health and growth.

With a new 10 Year Health Plan and a Government committed to scientific innovation as a major driver of economic growth, the opportunity is now. As the process of transitioning NHS England into the Department of Health and Social Care (DHSC) gets underway, the timing is ideal. Learning from the successes of NCDs in other major health conditions – such as cancer, cardiovascular disease and neurology – this new role would drive clinical and policy focus for rare diseases, improve access to and uptake of medicines and provide vital clinical advocacy at the highest levels of the rare disease community. With a goal of ensuring not only that life science research, drug development and translational medicine are optimised, but also of improving the understanding by decision-makers of the conditions and of scientific advancements in treatment. This would serve to ensure that patient care is underpinned by the very best clinical evidence and that patients benefit from fast and equitable access to the very best medicines.



# DRIVING CLINICAL LEADERSHIP FOR RARE DISEASES

The **NHS is a clinically led system**, relying on expertise to drive decision-making. Nowhere is the opportunity cost of a wrong decision potentially greater than whether to grant access to new medicines. Studies have shown that in health technology assessment (HTA) in rare disease, “clinical challenges relate to the scarce scientific literature and number of clinical experts available, where often little is known about the diseases’ epidemiology, natural history or best treatment pathways”.<sup>1</sup>

However, in rare diseases, expertise is often fragmented, in part because rare and complex conditions often necessitate multidisciplinary specialised care.<sup>2</sup> This expertise is typically also limited to just a handful of specialists and spread across national and international centres of excellence.

This results in several key challenges:

- **Lack of coordinated national leadership:** Unlike cancer, cardiovascular disease, or stroke – each with dedicated NCDs – rare diseases lack unified representation at the highest levels, which impacts on the UK’s ability to attract research and to provide access to the best care.
- **Clinical uncertainty in NICE appraisals:** Current perceptions of suboptimal evidence or bias already add uncertainty to the HTA process. The changes in highly specialised technology (HST) criteria will likely disadvantage rare disease interventions too.<sup>3</sup> Without a clear, consensus-driven clinical voice, NICE assessments of treatments for rare diseases will likely continue to suffer from insufficiently high expert input, potentially leading to greater uncertainty and suboptimal decision-making.<sup>4</sup>
- **Delays in diagnosis and access to care:** The ‘diagnostic odyssey’ for rare disease patients – already a source of great anguish for patients and unnecessary cost for the NHS – is prolonged due to **low awareness among general clinicians** and a lack of specialist input in local healthcare settings. There is an opportunity for greater coordination driven by stronger leadership.
- **Increasing demand for rare disease expertise:** Advances in **genomics, newborn screening, and precision medicine** are identifying more rare diseases, requiring enhanced clinical leadership to manage new diagnostic and treatment pathways. Greater scientific understanding brings advancement in treatment options, which presents both challenges and great opportunities for the NHS.

# CHAMPIONING THE VOICE OF CLINICIANS

The burden of undiagnosed rare diseases on the NHS is estimated at over £3.4 billion over 10 years, and this will only grow.<sup>5</sup> As such, helping patients get a final diagnosis faster was one of the 4 key commitments of the 2021 UK Rare Disease Framework.<sup>6</sup> A dedicated NCD would drive this ambition, helping to address fragmented rare disease services and to capitalise on advances in science and medicine.

The UK Government has signalled strong intent to **elevate scientific innovation and patient-centred healthcare**. The 2021 UK Rare Diseases Framework set out key priorities, and the 2024 England Action Plan called for better policy alignment. However, leadership remains diluted across multiple organisations, lacking a single, authoritative clinical voice.

A new Government with a focus on **harnessing life sciences and strengthening clinical leadership** – seen through the Labour Manifesto 2024 commitment to establishing a Royal College of Clinical Leadership – presents an opportunity to embed an NCD for Rare Diseases into the next iteration of the Rare Diseases Framework (2026). This will go some way towards the Government's stated intention of 'championing the voice of clinicians'.<sup>7</sup>

## THE SOLUTION: A National Clinical Director for rare diseases



We propose the creation of a National Clinical Director for Rare Diseases, reporting directly to the National Medical Director for the NHS. This role would ensure a strategic, coordinated, and expert-led approach to rare disease policy, commissioning, and service provision.

While each disease may be rare, overall, their presence in the population is less so, with estimates that 1 in 17 people in the UK will be affected by a rare condition at some point in their lives.

While there will be many and varied individual conditions, patient communities will have similar experiences and challenges, including in accessing cutting edge medical care.

# KEY RESPONSIBILITIES OF THE NCD

- 1 Improve quality and outcomes** by aligning clinical priorities with NHS service planning and commissioning.
- 2 Drive life sciences investment** in research & development and increase access for patients to commercial clinical trials.
- 3 Provide national clinical leadership**, coordinating expertise nationally and internationally to support decision-making. This could include high level leadership for the transition from NHS England to DHSC of groups such as rare disease collaborative networks (RCDNs) and the rare disease advisory group (RDAG), ensuring accountability for delivery of key commitments is not lost.
- 4 Guide NICE appraisals** by offering or coordinating expert clinical input to ensure fair and balanced assessments of new treatments. This might include liaising with international colleagues, particularly for conditions affecting very small patient populations, to gain from clinical experience overseas.
- 5 Champion patient-centred care**, enabling co-production with patients and the public to shape patient pathways and service delivery with a view to improving equitable access to diagnostics, treatment and care.
- 6 Promote health equity**, ensuring rare disease representation across all major health and care institutions, helping to advance integration and joined up thinking between health and social care settings.
- 7 Support workforce development**, creating tools and training to enhance knowledge of rare diseases across different healthcare settings.



# PROVEN IMPACT OF NCDs IN OTHER SPECIALITIES

The existing NCD model has demonstrated significant benefits in other major health areas:

- **Neurology NCD:** Established following a campaign by the Neurological Alliance,<sup>8</sup> it has driven the **Neurology Transformation Programme**, enhancing access to services and coordinating national expertise.<sup>9</sup>
- **Cancer NCD:** Led the development of **Cancer Healthcare Goals**, securing £22.5 million in Government funding and positioning the UK as a leader in cancer innovation.<sup>10</sup>
- **Cardiovascular Disease NCD:** Enabled a national focus on **prevention and treatment**, improving patient outcomes through coordinated policy and research efforts.<sup>11</sup>

Rare diseases share common challenges with these disciplines – **heterogenous patient populations, complex pathways, and the need for specialised expertise** – yet lack the **dedicated leadership** needed to drive improvements.

Patients with rare diseases are subject also to threats to progress in their diagnosis, treatment and care not only from the upheaval during the transition at national level, but from significant forthcoming changes at local systems level too. An NCD in rare could provide coordination and connection through the ecosystem down to Integrated Care Board (ICB) level.

## HIGH-PROFILE ADVOCACY AND MOMENTUM



**Lord David Cameron**, former Prime Minister, now chairs the **Oxford-Harrington Rare Disease Centre**, leading international efforts to develop **40 new rare disease treatments** in the next decade.<sup>12</sup>



**Dame Nicola Blackwood**, living with **Ehlers-Danlos Syndrome**, has been a vocal advocate for rare diseases and now chairs **Genomics England**, pushing forward advancements in rare disease diagnostics and treatments.<sup>13</sup>



The **Rare Autoimmune Rheumatic Disease Alliance (RAIRDA)** has called for a **Quality Statement for Rare Diseases**, galvanising widespread clinical and patient support.<sup>14</sup>

# NEXT STEPS: MAKING THE NCD FOR RARE DISEASES A REALITY

To drive this proposal forward, we recommend the following actions:

- **Undertake a short study** to validate the benefits of NCDs in other disciplines and project potential impact on rare diseases.
- **Engage the clinical and patient communities** to build consensus and gather support from professional societies, advocacy groups, and NHS leaders.
- **Define success metrics**, establishing how the NCD's impact on patient outcomes and healthcare services will be measured.
- **Secure policy commitment**, aiming for inclusion of an NCD for Rare Diseases in the 2026 UK Rare Diseases Framework.



## CONCLUSION: SEIZING THE OPPORTUNITY

The establishment of an NCD for Rare Diseases would be a **transformative step** for the NHS, ensuring rare disease patients receive the same clinical focus, investment, and strategic leadership as other major conditions. With the policy environment shifting towards greater clinical leadership, and rare disease innovation accelerating, now is the time to make this role a reality.

By learning from successful NCD models and leveraging the UK's leadership in genomics and rare disease research, this role would **drive lasting improvements in diagnosis, treatment, and patient care** – providing hope for the millions of people living with rare diseases.



If you have any questions or would like to discuss any of the issues raised in this report, please contact Jessica March, External Affairs Lead for Rare Disease at Biogen UK & Ireland, at [jessica.march@biogen.com](mailto:jessica.march@biogen.com).

## About Biogen

Founded in 1978, Biogen is a leading global biotechnology company that has pioneered multiple breakthrough innovations for people living with serious neurological diseases as well as related therapeutic adjacencies. Biogen has a portfolio of medicines and potential therapies across neurology, neuropsychiatry, specialised immunology and rare diseases and remains acutely focused on its purpose of serving humanity through science while advancing a healthier, more sustainable and equitable world.

To learn more, please visit [www.biogen.uk.com](http://www.biogen.uk.com).

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