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Hidden in Plain Sight: A Blueprint for Specialist Nursing

Developed by a UK Rare Disease Specialist Nursing Steering Committee comprising specialist nurses and patient organisations. Funded and convened by Alexion, AstraZeneca Rare Disease.

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- Juliette Buttimore
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Disclaimer

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To find out more, please contact:

Sam Briggs

Senior Manager, Corporate Affairs UK

UKcorporateaffairs@alexion.com

Alexion Pharma UK Ltd, 6th Floor, 2 Pancras Square, London, N1C 4AG

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Foreword

Throughout my engagement with the rare disease community, I have been reminded time and again that good care depends not only on clinical expertise, but on how effectively services work together around the needs of people living with rare disease and their families.

For many, care spans multiple specialties, organisations and stages of life, requiring services that provide continuity and personalised care across traditional boundaries. It is against this reality that the current policy moment and the opportunity it presents to strengthen joined-up, patient-centred care should be understood.

The publication of the National Institute for Health and Care Excellence (NICE) Quality Standard for Rare Diseases has, for the first time, formally endorsed what good rare disease care should look like, while the conclusion of the UK Rare Diseases Framework prompts us to consider what comes next aligned to this quality standard.

A new government focused on improving outcomes in every postcode where every place and every person matters also brings fresh opportunities to consider how we deliver more joined-up, patient-centred care across the NHS. At the same time, the NHS Long Term Workforce Plan has reinforced the need to invest in, retain and grow the specialist workforce required to meet increasingly complex patient needs.

As this blueprint demonstrates, rare disease Clinical Nurse Specialists (CNSs) exemplify the kind of coordinated, person-centred care that health policy has long sought to achieve. Every day, they work across organisational boundaries, connect services that were not designed to work together, support families through complex treatment journeys and provide continuity for people navigating often fragmented systems.

What I particularly welcome about this blueprint is that it has been shaped by those closest to rare disease care. Drawing on the experiences of CNSs, patient organisations and people living with rare disease, it offers a practical perspective on both the opportunities and barriers within the current system.

As policymakers consider the future direction of rare disease policy, it is essential that decisions are informed not only by national strategies and frameworks, but by the experience of those delivering and receiving care on the ground. The perspectives captured in this blueprint provide an important contribution to that conversation and a valuable foundation for future action.



Margaret Ritchie,
Baroness Ritchie of Downpatrick

Executive summary

Rare disease care is inherently complex.

People living with life-long and chronic rare conditions often navigate multiple services, specialists and settings over the course of their lives as their clinical and wider health needs change, frequently coordinating their own care and re-explaining their condition as they go. The result is a care journey that can feel fragmented, inconsistent and difficult to understand.

While individual conditions may differ, people living with a rare disease consistently rely on the same core specialist nursing functions: coordination, triage, continuity and holistic support, underpinned by expertise in managing rare and complex conditions.

This creates a clear case for a two-part workforce model. Where condition-specific specialist nurses are already in place, their roles should be clearly defined, properly resourced and sustained. Where a dedicated condition-specific post is not viable, services should establish pan-rare specialist roles that support people across several rare conditions, while working with condition-specific clinicians and patient organisations where deeper expertise is required. This ensures that people do not go without essential coordination, continuity and specialist support.

Cross-condition models are achievable. An independent Sheffield Hallam University evaluation of the Roald Dahl Specialist Nurses, a workforce of over 200 nurses supporting more than 40,000 children across a range of rare and complex conditions, found families rated their care an average of 9.47 out of 10, with the role improving coordination, family support and efficient use of NHS resources.[1]

Despite the positive impact of CNSs on patients and their families, there is a clear disconnect between professionals' perceptions of provision and patients' experiences. While 35% of healthcare professionals believe patients have access to a care coordinator, only 12% of adults with a rare disease report having one, and just 2% report having access to the full combination of a care coordinator, specialist centre and care plan.[2] This falls well short of NICE's expectation that every person with a rare disease should have access to a named healthcare professional responsible for coordinating their care.

The publication of the NICE Quality Standard for Rare Diseases presents an opportunity to close this gap and others commonly identified throughout many rare disease pathways. It is especially important as NICE does not routinely develop clinical guidelines for individual rare diseases, instead focusing on common challenges cutting across conditions. The quality standard therefore provides a system-wide benchmark through its eight Quality Statements, setting out what good care should look like, from suspected diagnosis through to access to treatment.[3] The task now is to deliver that standard consistently across the NHS.

This blueprint demonstrates that where they exist, CNSs already deliver much of the care the standard requires, but without nationally defined roles, responsibilities and models of provision for specialist nursing, the NHS will remain limited in its ability to identify capacity, plan services and address persisting care gaps.

The blueprint proposes seven recommendations spanning **national**, **system** and **service levels** with measurable indicators to support implementation and accountability. These are accompanied by five enabling conditions that are essential to sustain specialist nursing, including workforce visibility, multidisciplinary support, administrative infrastructure, analytical capability and professional development. Together, these form the foundations needed to deliver high-quality rare disease care.

Summary of recommendations

The blueprint proposes seven recommendations spanning national, system and service levels with measurable indicators to support implementation and accountability.

National



Make the rare disease CNS workforce visible within national workforce planning

Establish a national standard for identifying CNS posts supporting people with rare diseases. In the absence of a dedicated rare disease workforce category, NHS workforce data for complex, specialised and highly specialised services should be used as an initial metric, supported by consistent role descriptors and service-level validation.

Measure

Proportion of CNS posts within complex, specialised and highly specialised services that can be identified by service area, role and population supported, with local validation of posts providing rare disease care.



Strengthen national approaches to measurement and accountability

Develop a national approach to measuring specialist nursing capacity, access and contribution to rare disease care. This should combine workforce and service data with a regular national measure of rare disease patient experience. These measures should enable variation in provision and workforce gaps to be identified and addressed.

Measure

A defined national set of rare disease specialist nursing indicators is incorporated into routine reporting, alongside a regular national measure of patient experience.

Regional/Local Health Systems



Require specialist nursing within rare disease service specifications

All existing and future rare disease service specifications should define the role and required capacity of specialist nurses within the multidisciplinary team. This should include responsibility for care coordination, treatment support, patient education and holistic care.



Measure

Proportion of existing and newly developed rare disease service specifications that explicitly define specialist nursing provision, responsibilities and supporting multidisciplinary infrastructure.



Ensure equitable access to specialist nursing

Work towards needs-based workforce planning for specialist nursing by setting a minimum expectation of access to specialist nursing support regardless of condition, geography or commissioning arrangement, with particular focus on care coordination, treatment support and transition between paediatric and adult services.



Measure

Proportion of people with a rare condition with access to a named CNS coordinator, and variation in access by region and condition.



Formally recognise CNSs as named care coordinators where appropriate

Where CNSs already fulfil the functions set out in NICE Quality Statement 5, organisations should formally recognise and support them as the named healthcare professional responsible for coordinating care.

Measure

Proportion of relevant services that formally designate the CNS as named care coordinator within the care pathway and communicate this to patients and the wider MDT.



Define, protect and support the CNS role

Organisations should use clear job descriptions, transparent service organograms and formal job planning to define the scope of the CNS role and prevent wider service gaps from displacing specialist nursing activity. Job plans should protect time for direct patient care, care coordination, service improvement, research, leadership and continuing professional development.

Measure

Proportion of rare disease CNSs with an agreed job description and formal job plan covering the full scope of the role.

Proportion with protected CPD, leadership, research or service-improvement time.

Proportion of services with defined administrative support and transparent allocation of responsibilities across the MDT.



Ensure specialist nursing time is used effectively

Provide appropriate administrative support and access to multidisciplinary teams so specialist nursing expertise is focused on activities requiring clinical judgement, coordination and specialist knowledge.

Measure

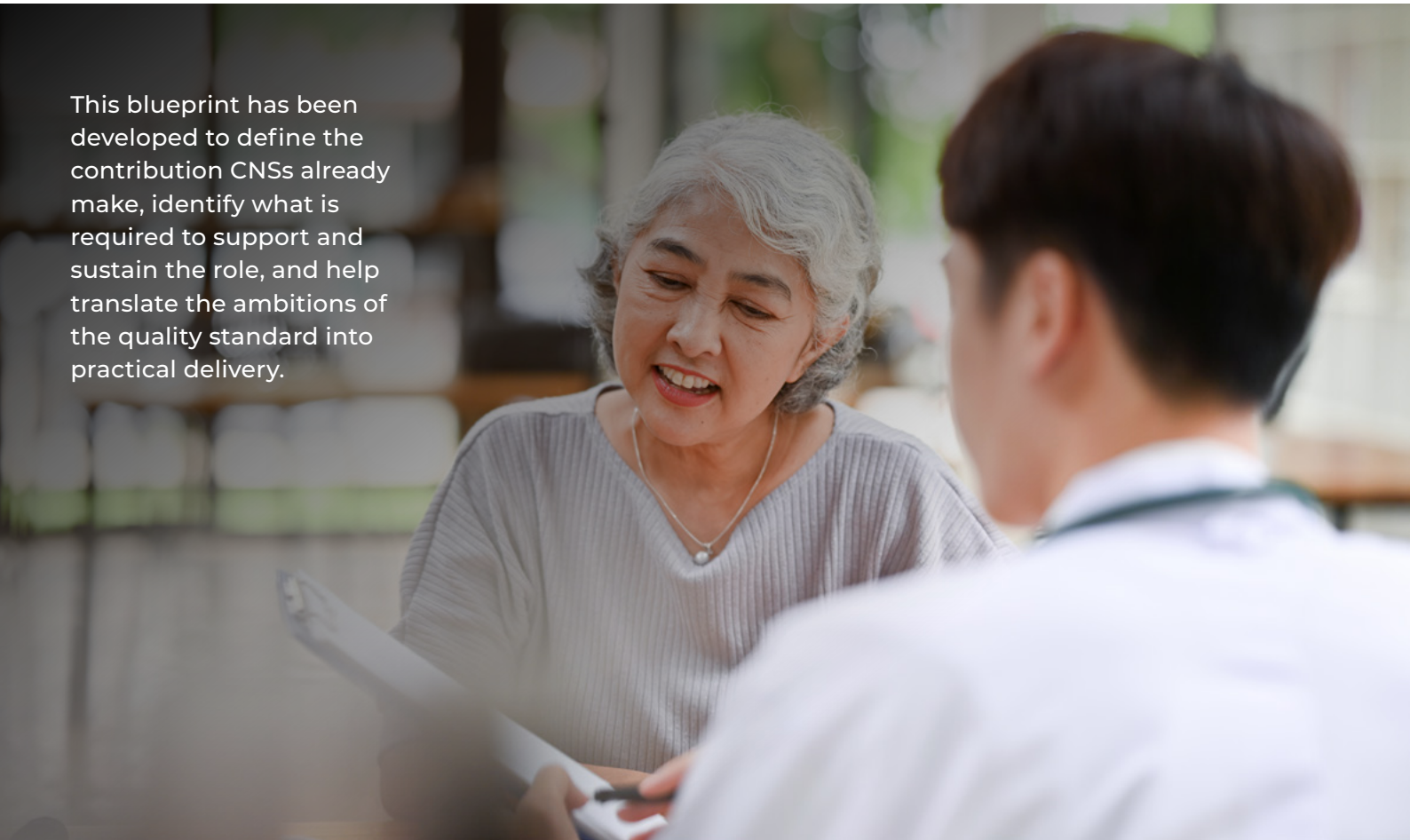
Minimum admin-to-CNS ratio within specialist rare disease services, incorporated into service specifications.

01. The delivery moment: NICE Quality Standard

The publication of the NICE Quality Standard for Rare Diseases marks an important shift in the policy environment. For the first time, there is a clear and measurable system-adopted description that provides a system-wide benchmark for rare disease care.

This comes at a period of significant change across the NHS. As commissioning responsibilities continue to evolve, and national and system-level infrastructure becomes more limited, clear standards and consistent definitions of good care become increasingly important. The quality standard provides guidance for commissioners, providers and system leaders, helping ensure implementation does not become dependent on local interpretation or service history.

It also arrives at a time when workforce planning is receiving renewed national attention. While CNSs are recognised within national workforce planning in areas such as cancer in the Long Term Workforce Plan, there remains no equivalent recognition of the rare disease CNS workforce.[4] The quality standard provides an important opportunity to address that and this blueprint has been developed in response to that opportunity: to define the contribution CNSs already make, identify what is required to support and sustain the role, and help translate the ambitions of the quality standard into practical delivery.



02. Who are Clinical Nurse Specialists?

Clinical Nurse Specialists (CNSs) have been part of the NHS workforce for decades.

In rare disease services, CNSs are senior nurses with specialist expertise in a condition, service or patient population. The role is a salaried, advanced nursing position, based predominantly in NHS hospitals, with senior nurses who hold specialist expertise in a condition, a service or a patient group. Most are educated to degree level or working towards a master's-level qualification; many sit at Agenda for Change Bands 6 to 7, with a smaller number of nurse consultants at Band 8a or 8b. Across NHS specialised services, specialist nurses are recognised as core members of multidisciplinary teams. For example, NHS England service specifications for specialised rheumatology and cystic fibrosis services explicitly identify specialist nurses within the MDT, reflecting their role in coordinating care, supporting treatment pathways and providing continuity for people with complex conditions.[5,6]

There is no national specification for the role, and a systematic audit of 100 CNS job descriptions across the UK found considerable variation in titles, pay bands, entry requirements and post-registration qualifications. However, the audit identified shared functions that provide a basis for a pan-rare workforce argument, without suggesting that all rare diseases, services or nursing roles are the same[7]:

In rare disease, that variability is compounded by a gap in access. Where a CNS exists, provision depends heavily on how the role has been set up locally rather than on any agreed standard, and for many people living with rare disease, no dedicated specialist nurse exists at all. The result is fragmented and inconsistent access, with many left without any specialist nursing support.

1. **Clinical practice**
   Direct patient care in a defined speciality.

2. **Leadership and management**
   Quality improvement, safety, supervision.

3. **Education**
   Teaching colleagues, people with rare disease and their families.

4. **Research and improvement**
   Evidence-based practice, contribution to research.

5. **Professional and ethical practice**
   Advocacy, accountability, NMC Code adherence.

2.1 The role in practice: five core responsibilities

Across services, CNSs described maintaining oversight of the patient journey, coordinating care and ensuring that concerns are escalated appropriately. Their contribution can be understood through five core responsibilities:

1. Coordinating multidisciplinary care:

CNSs act as the connecting point between hospital specialists, primary care, diagnostics, pharmacy, community services and patient organisations. They maintain oversight across the pathway, ensuring information is shared appropriately and helping prevent people from becoming lost between services.



2. Triaging clinical concerns:

People with rare disease and families frequently contact CNSs when concerns arise. Nurses assess the urgency of issues, provide practical advice, identify situations requiring escalation and help avoid unnecessary use of emergency or specialist services.



3. Supporting treatment pathways:

CNSs help ensure people with rare disease can access treatment safely and effectively, coordinating appointments, monitoring ongoing needs, troubleshooting barriers and helping services adapt to changing clinical circumstances.



4. Educating healthcare professionals:

Rare disease often sits outside routine clinical experience. CNSs support colleagues across healthcare settings by sharing expertise, providing education and building confidence in managing rare disease.



5. Anticipating and escalating risk:

CNSs combine detailed disease-specific knowledge with a long-term understanding each person's circumstances. This enables them to identify deterioration, emerging risks or unmet needs early, whilst also recognising issues that can be managed safely without additional intervention.



2.2 When gaps in care widen the CNS role

CNSs often take on responsibilities beyond their intended clinical remit because essential support is missing elsewhere in the care pathway. The example below shows how this happens in practice. It demonstrates both the value of having a specialist nurse who can see the whole picture and the risk of relying on one individual to compensate for gaps across multiple services.

How a specialist nurse held a complex care journey together

1.

An unrecognised rare condition:

A child with a life-threatening tumour was referred after the CNS's specialist teaching helped a local clinician recognise the need for expert support.



2.

Connecting a fragmented pathway:

The CNS became the family's consistent point of contact, coordinating care across three hospitals and more than ten specialties.



3.

Escalating unmet needs:

During a complex admission, the CNS secured psychological support despite the child not meeting the usual criteria and supported multidisciplinary discharge planning.



This example shows the breadth of value CNSs can provide, but it should not be treated as an expectation that one nurse will absorb every unmet need around a family. As responsibilities expand, services must clearly define the CNS role and provide the multidisciplinary, administrative and community support needed around it. Understanding, measuring and supporting this contribution is the first step towards sustaining it.

4.

Building local knowledge and capacity:

The CNS trained more than 20 local paediatricians. This contributed to a consultant taking a lead for the condition locally and work to identify affected children and improve clinical guidance.



Supporting the whole family:

The CNS trained the child's school, supported the sibling's school, appealed a rejected disability benefit application and prepared reports for education and continuing-care assessments.



5.

6.



Bringing care closer to home:

Community blood monitoring was arranged to reduce travel and make ongoing care more manageable for the family.

03. Delivering the Quality Standard: what CNSs make possible

CNSs already deliver many of the responsibilities within the Rare Disease Quality Standard, often providing the continuity, coordination and expertise that underpin high-quality rare disease care.

This section sets out their contribution to each quality statement, drawing on published evidence and insights from the Specialist Nursing Steering Committee.

Evidence specific to rare disease specialist nursing remains sparse, reflecting that much of the role's value is generated through preventative interventions, coordination activities and patient support that are not routinely recorded or measured. Consequently, specialist nursing impact is often under-recognised despite being critical to service delivery. The examples presented throughout this section demonstrate capabilities that extend beyond individual conditions and services, offering practical illustrations of how specialist nursing expertise can be applied, adapted and scaled across rare disease care.





3.1 Statement 1:
Referral for investigation and treatment

In recognition of the diagnostic odyssey, the first Statement calls for prompt referral to consultant-led services and access to confirmatory testing to support timely treatment decisions. Whilst CNSs are rarely responsible for the original referral itself, they often become central to what happens next.

CNSs help coordinate confirmatory investigations, support treatment initiation, explain complex findings and help people with rare disease and their families navigate the transition from diagnosis into ongoing care.

> A child referred following genetic diagnosis was expected to wait two months for specialist review, followed by a further delay before treatment could begin. Through direct coordination between local and specialist teams, the CNS expedited testing and arranged an urgent appointment, reducing the time to specialist review to two weeks and enabling treatment to start within four weeks of referral. The intervention demonstrates the role specialist nurses play in turning diagnosis into timely access to care.

This contribution is particularly important within genetic and genomic services, where diagnosis is rarely a single event, but instead involves investigation, interpretation, further review and adaptation over time. Working alongside consultant-led multidisciplinary teams, CNSs help identify people suitable for genomic testing, guide individuals and families through testing and the interpretation of results and support them from diagnosis into ongoing care.[8]

The value is already visible in practice at Guy's and St Thomas' nurse-led genomics service, which facilitates expedited genetic testing to identify inherited conditions, meaning people with rare disease and their families can be tested faster and receive continued specialist support even when seen at their local hospital.[9]

3.2 Statement 2:
Undiagnosed conditions

The second Quality Standard recognises people with a suspected rare disease who remain undiagnosed as a distinct patient group. Around 6,000 children are born in the UK each year with a genetic condition so rare it does not yet have a name, and roughly half of all children undergoing genetic testing in the UK do not receive a confirmed diagnosis.[10,11]

The quality standard makes it clear that these individuals should not be left without direction once initial investigations have been exhausted, but should have ongoing support, review and a documented care plan. For many people living without a diagnosis, CNSs become the professional who helps maintain continuity through a long and uncertain diagnostic journey. This role is especially important because the absence of a diagnosis can make it harder for people with rare disease to access condition-specific information, peer support or a clearly defined pathway.

The Roald Dahl SWAN (Syndromes Without A Name) CNS at Great Ormond Street Hospital, the first post of its kind in the UK, supported 40 undiagnosed families in its first year. On average, the children were under the care of seven different hospital teams. The CNS coordinated appointments to streamline visits, helped families understand and process complex clinical information, and provided ongoing emotional and social support, with feedback from families and clinicians described as universally positive.[12]

The Cardiff and Vale SWAN service also showed that coordinated support for people living without a diagnosis was associated with improved patient experience, greater self-management and a large reduction in secondary care contact days.[13] There is evidence from other conditions that the effective use of clinical nurse specialists can help reduce costs by minimising the need for inpatient care or emergency admissions.[5]

3.3 Statement 3:
Information provision

The third quality standard recognises that people living with a rare disease need timely access to accurate information to understand their diagnosis, navigate services and make informed decisions about their care.

There is an acute inequity of access to information in rare disease compared with common, population health conditions. In a UK-wide survey of over 1,300 people living with rare autoimmune rheumatic conditions, only 16% felt they had enough access to information and support.[14] Access to specialist nursing support was associated with substantially better experiences: 79% of respondents who had access to a specialist nurse felt they had enough information and support, compared with 58% of those without access to a specialist nurse.[14]

Where information is unclear or inconsistent, it can increase anxiety and reduce people's ability to participate in decisions about their care.[15]

In rare diseases, this challenge is amplified by the difficulty of finding reliable, condition-specific information,[16] with 47% of patients feeling the support they receive is generic rather than tailored to their circumstances.[17]

In the absence of widely available, high-quality, condition-specific information, CNSs can provide deep subject matter expertise and signpost support. Steering Committee members described spending substantial time with people with rare diseases and their families outside formal consultant appointments, in some cases sitting with individuals for more than an hour to talk through diagnoses, treatment options, anxieties and the practical implications of living with rare disease. By doing so, they reduce avoidable follow-up queries, support patient confidence and release consultant time for decisions that require senior medical input.

CNSs frequently work with patient organisations to co-design accessible information resources and patient-friendly educational materials. In rare diseases, where lived experience, peer support and practical guidance can be critical, this partnership between specialist services and patient organisations is an important part of high-quality information provision.



3.4 Statement 4: **Shared decision-making**

The fourth quality standard recognises the need for people living with rare diseases to be active participants in decisions about their care throughout the course of their condition. Decisions about rare disease care are rarely made at a single point in time – this is particularly true for lifelong and chronic conditions. Management plans evolve as symptoms change, treatment options develop, family circumstances shift and people come to terms with the long-term implications of their condition.

CNSs are what make shared decision-making possible over that long arc. Because they hold sustained relationships with patients and families, often over years, they can tell when a decision has not been fully understood, when circumstances have shifted, or when a conversation needs revisiting. Families contact them between appointments to query medication changes or clarify options, avoiding delays and preventing people from remaining on the wrong course while they wait for the next clinic.



3.5 Statement 5: **Named healthcare professional**

Of all the statements, care coordination is most closely aligned to the CNS role. Quality Statement 5 requires people living with rare disease to have access to a named healthcare professional responsible for coordinating their care.

Steering Committee participants consistently described fulfilling this function. CNSs connect services, coordinate care across organisational boundaries, triage concerns and apply specialist expertise to identify issues requiring escalation.

In doing so, they help patients navigate complex pathways, direct them to the right support and reduce avoidable appointments, investigations and emergency care use. For many people living with rare conditions, CNSs are the only professionals with oversight of the whole patient journey.

Evidence from other disease areas reinforces the importance of this function. Analysis linking the National Cancer Patient Experience Survey with cancer registry data found poorer outcomes among patients who reported not being given the name of a CNS, highlighting the value of continuity and coordinated support within complex pathways.[18]

Condition-specific services demonstrate what becomes possible when specialist nursing is embedded within a care pathway. In cystic fibrosis, specialist nurses are a recognised component of accredited multidisciplinary teams, reflecting the central importance of dedicated care coordination.

> A family was referred to the specialist nurse through a genetic counsellor, after the mother – who lives with a dominantly inherited rare condition – became pregnant. Working within clinical genomics, the nurse supported planning from before birth: a neonatal alert was established and cord-blood sampling arranged at delivery, ensuring timely testing in line with the parents' wishes.
>
> Recognising that the mother's condition carried pregnancy-related risks unfamiliar to local teams, the nurse wrote to the wider obstetric team to flag them directly. This ensured the pregnancy was managed safely and that appropriate postnatal plans were in place before the birth. These risks would otherwise have gone unrecognised locally.
>
> The baby inherited the condition, and the nurse delivered the news jointly with the genetic counsellor. While the counsellor's role ended at diagnosis, the nurse became the family's consistent point of contact to this day. In that time, they have coordinated a referral to physiotherapy for developmental concerns linked to the condition and written to local paediatric teams with clinical guidelines and surveillance requirements to ensure the child is followed up appropriately despite limited local expertise. The family continues to turn to the nurse for help navigating the system whenever local services are uncertain about how to manage the child's rare disease.

During COVID-19 service redesign at a large adult cystic fibrosis centre, CNSs managed 1,187 patient contacts in a single month, using telephone triage to maintain continuity of care and identify those requiring face-to-face review.[19]

However, access to specialist nursing support varies considerably between conditions, regions and commissioning arrangements. For conditions that lack the scale or commissioning arrangements to sustain a dedicated condition-specific CNS workforce, pan-rare specialist nursing roles may offer an alternative route to delivering care coordination, while drawing on condition-specific clinicians and patient organisations when deeper expertise is required.

> “Many people with aHUS do not have access to a Clinical Nurse Specialist or other named healthcare professional responsible for coordinating their care. In the absence of dedicated specialist nursing support, coordination responsibilities often fall to other professionals, such as renal pharmacists, whose expertise lies elsewhere. While these workarounds help patients navigate the system, they are difficult to sustain and increase the risk of individuals becoming lost between services. The experience demonstrates that effective care coordination requires dedicated capacity, not informal arrangements.”



3.6 Statement 6: **Holistic support**

Rare diseases affect far more than physical health alone, requiring support that reflects the wider impact of living with a complex long-term condition. The sixth quality standard recognises this need to assess physical, psychological, emotional and social needs alongside clinical management.

As covered earlier in this Blueprint, CNSs routinely identify psychological distress, family pressures, financial concerns and practical barriers to care. For example, 62% of people affected, either directly or indirectly, by rare conditions affecting the bone marrow, want more information on mental wellbeing and 39% of those not currently speaking to a psychologist, therapist, or counsellor, feel that speaking to one would increase their quality of life.[20]

In the absence of embedded psychological or social support, CNSs often become the professionals whom people with rare disease and their families turn to first. This reflects the trust placed in CNSs but also exposes a wider system problem. Holistic care cannot rest on CNSs alone if the wider multidisciplinary infrastructure is not available.

Holistic care extends beyond psychological support. People with rare disease and their families often need support across paediatric to adult transition, education, employment, social care and welfare services, yet access to this wider infrastructure remains inconsistent and heavily dependent on local service design.



3.7 Statement 7:
Access to treatment

The seventh quality standard recognises that access to treatment must be supported by appropriate monitoring and ongoing clinical review. CNSs provide much of the infrastructure that makes access to treatment possible in practice. They support treatment initiation, monitor safety and effectiveness, identify barriers to adherence and provide a trusted point of contact when concerns arise. This is particularly important in rare disease care, where treatments may be highly specialised and require careful monitoring and clinical oversight, while people may also need support to manage treatment alongside complex wider needs.

Access to treatment should therefore be understood as more than the availability of a medicine, procedure or clinical appointment. People also need the specialist nursing capacity required to start treatment safely, manage complications, and access timely advice when problems arise.

In a study of Roald Dahl Specialist Nurses supporting children with serious and rare long-term conditions, timely contact with the nurse averted consultant appointments for 52.7% of families on at least one occasion, prevented urgent GP appointments for 40%, averted A&E visits for 42.3%, and prevented at least one emergency hospital admission for 37.5% of families in the previous year. These “open access” models, often delivered through a nurse-led telephone line, reduce demand on emergency and urgent care precisely when system capacity is most stretched.[1]

Avoided admissions, earlier intervention and improved coordination may influence outcomes, but are not always directly attributable to a single activity or intervention.



3.8 Statement 8:
Clinical research

For about 95% of rare diseases, there is still no approved disease-specific treatment.[21] Generating robust clinical evidence depends on participation from small and geographically dispersed patient populations. The eighth Quality Standard highlights the importance of ensuring that those individuals and their families are aware of relevant research opportunities and supported to participate where appropriate.

Through the CNSs’ detailed knowledge of patient populations and long-term relationships with people and their families, they are well placed to identify potentially eligible individuals, support conversations around participation and coordinate involvement alongside routine care.

CNSs can also help ensure research participation is discussed in a way that is meaningful and appropriate: they can explain what taking part may involve, identify practical barriers, support follow-up and help families weigh research participation alongside treatment, work, school, caring responsibilities and wider life pressures. NHS England’s Nursing and Midwifery

Genomics Framework identifies nurses as central to enabling access to genomic testing and research, and government policy is now explicitly positioning nurses to lead and deliver research into rare and inherited conditions.[8]

Recognising the CNS role as a core component of research delivery infrastructure would help ensure more people with rare disease are able to access research opportunities and contribute to the development of future treatments.

> “As our service expanded to cover Scotland, we used part of the additional funding to recruit dedicated administrative support rather than investing solely in nursing posts. This created a clearer division of responsibilities, improved efficiency and released specialist nursing time for direct patient care, care coordination and service improvement. The experience reinforced that administrative support is not a luxury within specialist services. It is essential infrastructure that allows specialist nurses to focus on the activities that deliver the greatest value for patients and the NHS.”



04. The enablers: what specialist nursing needs to work



Administrative support

One of the most consistent concerns raised during the development of this blueprint was the reduction or removal of administrative support within specialist services. Tasks such as appointment management, referral coordination, correspondence and service administration increasingly fall to CNSs when dedicated support is unavailable. Every hour a CNS spends on administration is an hour of clinical expertise lost to the service.

Administrative support is a prerequisite for an efficient service, and appropriately trained administrators perform many of these responsibilities more effectively and at a lower cost than senior clinical staff.



Access to embedded MDT roles

Access to psychology, specialist pharmacists, social work and welfare support remains inconsistent across rare disease services, as highlighted in England's Rare Diseases Action Plan 2025. For complex and lifelong rare conditions, care needs change over time and often extend beyond clinical management alone.

Delivering the NICE Quality Standard therefore depends not only on access to specialist nursing, but on access to the wider multidisciplinary workforce needed to support physical, psychological, emotional and social needs. Where that team is incomplete, the responsibilities fall to the CNS, which is neither sustainable nor an adequate substitute for specialist expertise. Variation in access to these roles is a useful indicator of where implementation is falling short.



Data and analytics capability

The quality standard places significant weight on measurable outcomes and accountability, yet much of the value generated by CNSs remains difficult to capture.

Demonstrating impact is challenging because many specialist nursing activities are preventative in nature and therefore invisible. The scale of the invisible workload is striking. One CNS, working 22.5 hours per week as the sole specialist nurse for a regional population of 4.5 million people, reported supporting approximately 600 patients, receiving or sending around 680 patient-related emails and managing 265 patient-related phone contacts each month. While these interactions represent a substantial proportion of care coordination, triage and patient support activity, they are rarely captured in routine performance measures.[22]

This is compounded by the decline of dedicated data and service management support. Where analytical capacity is unavailable, responsibility for audit, reporting and data collection can fall to clinical staff, further stretching already limited resources.

At both service and national level, stronger analytical capability will be needed to demonstrate impact and monitor implementation. The availability of dedicated data and analytical support is likely to be an important indicator of whether services have the infrastructure required to evidence outcomes and support improvement.



Protected development time and CPD

Delivering high-quality rare disease care depends on clinical expertise that develops over years. Several Steering Committee members highlighted a lack of clear career progression routes within rare disease specialist nursing. This can result in highly experienced nurses leaving services to progress professionally, taking with them expertise and professional networks that may have taken years to develop. Workforce sustainability therefore depends on education and CPD, as well as on creating viable long-term career pathways for CNSs. Many nurses also reported having no formal job plan, limiting opportunities to protect time for professional development.

Protected time for learning and development should therefore be recognised as a core part of the role. As genomics, advanced therapies and increasingly specialised pathways become embedded within rare disease care, sustained investment in specialist expertise will become increasingly important. The complex and overlapping nature of some rare conditions means that knowledge of one condition alone may not be sufficient. CNSs may also need cross-cutting clinical knowledge of the underlying pathophysiology. Access to CPD and specialist development opportunities should form part of how organisations assess whether future workforce capability is being maintained.



Workforce visibility in national planning

Of all the barriers identified, visibility may be the most fundamental.

Rare disease CNSs are not consistently identifiable within national workforce datasets, service specifications or workforce plans. This limits understanding of workforce capacity, distribution, vacancy rates and future requirements across rare disease services.

As a first step, national workforce planning should use CNS posts within complex, specialised and highly specialised services as a proxy population, supported by standardised role descriptors and service-level validation to identify posts. This will not capture every rare disease CNS from the outset, but it would create a measurable baseline from which visibility and data quality can improve.

Greater visibility would support service planning, monitoring of the NICE Quality Standard and identification of inequities in provision. Over time, the proxy should be refined as definitions and coding improve.

05. Conclusion and call to action

The NICE Rare Disease Quality Standard has defined what good rare disease care should look like. The challenge now is to make that vision deliverable.

This blueprint demonstrates that specialist nursing is not a peripheral component of rare disease care, but one of the key enablers of the quality standard itself. Yet access to specialist nursing remains uneven and too often depends on condition, postcode or historic commissioning arrangements.

Where condition-specific CNS roles exist, they should be protected, clearly defined and sustainably resourced. Where dedicated posts are not viable, pan-rare specialist nursing models should provide the coordination, expertise and continuity that would otherwise be absent. No person living with a rare condition should be left without access to specialist nursing support because of where they live, what condition they have or how local services are organised.

Achieving this requires more than additional posts. It requires investment in the foundations that allow CNSs to work effectively: administrative and analytical support, integration within multidisciplinary teams, protected time for professional development, formal job planning, and visibility within national workforce planning and data systems. Without these enablers, the NHS will continue to rely on individual nurses absorbing unmet need elsewhere in the system, limiting the impact and sustainability of the role.

As the UK Rare Disease Framework reaches its conclusion and policymakers consider the next phase of rare disease policy, there is an opportunity to move from aspiration to implementation. Strengthening and expanding specialist nursing capacity offers one of the most practical and immediately deliverable ways to improve the experience of people living with rare conditions while supporting more coordinated and efficient care.



This blueprint demonstrates that specialist nursing is not a peripheral component of rare disease care, but one of the key enablers of the quality standard itself.

06. Appendix

6.1 How the Blueprint was developed

Alexion, AstraZeneca Rare Disease brought together senior Clinical Nurse Specialists and Patient Advocacy Groups together to better understand the contribution of CNSs and identify what is required to support and sustain their role. The aim was to provide a practical framework for delivering existing policy ambitions through greater recognition and support for specialist nursing. The blueprint was developed in three stages.

In 2025, Alexion, AstraZeneca Rare Disease convened the Rare Disease Patient Council, bringing together representatives from 12 patient organisations from across the UK to discuss priorities for improving rare disease care. Specialist nurse provision emerged as a particular priority, highlighting the need for a deeper exploration of how specialist nursing is delivered across rare disease services.

Following this, a series of qualitative scoping interviews was conducted by Alexion, AstraZeneca Rare Disease with specialist nurses and nurse leaders working across a range of rare disease services to understand how specialist nursing is delivered in practice, examining differences in service models, workforce pressures, barriers to expansion and examples of effective provision from across the UK.

A multidisciplinary Steering Committee comprising specialist nurses, nurse leaders and experts from across the rare disease community was convened. The Rare Disease Specialist Nursing Steering Committee played a critical role in testing emerging findings, validating areas of consensus and challenge, and ensuring that recommendations reflected the realities of delivering rare disease care within the NHS. Rather than representing a single specialty or service, the group was intentionally established to capture perspectives from across the rare disease landscape and identify themes that cut across conditions.

These insights were considered alongside national frameworks and guidance, including the UK Rare Diseases Framework, successive Action Plans and the NICE Quality Standard for Rare Diseases. The review also drew on specialist nursing workforce evidence, competency frameworks, service specifications, role profiles and lessons from more established specialist nursing workforces, including cancer care.

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