National Institute for Health and Care Excellence

Rare Diseases

Consultation on draft quality standard – deadline for comments 5pm on 20 January 2026

Please email your completed form to: QualityStandards@nice.org.uk

Please read the checklist for submitting comments at the end of this form. We cannot accept forms that are not filled in correctly.

Use the form to comment on the content of the quality standard (i.e. the statements and other sections e.g. rationale, measures etc.), as well as answer the following questions:

1. Does this draft quality standard accurately reflect the key areas for quality improvement?

2. Can data for the proposed quality measures be collected locally? Please include in your answer any data sources that can be used or reasons why data cannot be collected.

3. Do you think each of the statements in this draft quality standard would be achievable by local services given the net resources needed to deliver them? Please describe any resource requirements that you think would be necessary for any statement. Please describe any potential cost savings or opportunities for disinvestment.

4. Please provide your comments on the equality and health inequalities assessment (EHIA) and the equality and diversity considerations section for each quality statement. Please confirm any issues that have been missed and how they can be addressed by healthcare services and practitioners.

Implementing NICE guidelines

5. What are the challenges to implementing the NICE guidance underpinning this quality standard? Please say why and for whom. Please include any suggestions that could help users overcome these challenges (for example, existing practical resources or national initiatives).

Organisation details

Organisation name (if you are responding as an individual rather than a registered stakeholder please leave blank)	**Gene People**
Disclosure Please disclose any past or current, direct or indirect links to, or funding from, the tobacco industry.	
Name of person completing form	**Samantha Barber**
Supporting the quality standard Would your organisation like to express an interest in formally supporting this quality standard? More information.	**Yes**

Comments on the draft quality standard

Comment number	**Statement or question number** Or 'general' for comments on the whole document	**Comments** Insert each comment in a new row. Do not paste other tables into this table because your comments could get lost – type directly into this table.
Example 1	*Statement 1*	*This statement may be hard to measure because…*
1	General	Gene People, as a representative of the rare disease community, broadly supports the proposed quality standards and welcomes the focus on improving care and experience for people living with rare conditions, their families and carers. However, the real test of impact will be in implementation and action. Without clear accountability, consistent delivery and meaningful monitoring, the standards risk remaining aspirational. We stand alongside other rare disease voices, including Genetic Alliance UK and the Specialised Healthcare Alliance, in calling for a strong focus on turning these commitments into practical, measurable improvements across the system.

2	Question 1	Yes. If implemented effectively, this quality standard could improve outcomes and experiences for people living with rare diseases, their families and carers. It addresses the right priorities, including timely diagnosis, coordinated care, access to clear information, holistic support and equitable access to treatment and research. The focus on the whole care pathway, including people who remain undiagnosed, reflects lived experience well. However, its success will depend on implementation and sufficient resourcing at the local level; without this, the benefits will not be realised in practice.
3	Question 2	This question is outside our area of expertise.
4	Question 3	This question is outside our area of expertise.
5	Question 4	The Equality and Health Inequalities Assessment is high-level, and we see scope to increase specificity. We suggest revisiting the following: • **Socioeconomic status and deprivation:** People from deprived socio-economic backgrounds also experience lower health literacy rates, which can negatively affect their ability to advocate for themselves and actively participate in health decision-making (NHS Briefing: 4b_Health_Literacy-Briefing.pdf) • **Inclusion health and vulnerable groups:** Gypsy, Roma and Traveller communities are heavily disadvantaged in the healthcare system - https://www.gypsy-traveller.org/wp-content/uploads/2022/11/Briefing_Health-inequalities-experienced-by-Gypsies-and-Travellers-in-England.pdf. This is specifically relevant when it intersects with rare disease diagnosis/treatment/monitoring. The quality standard would also be improved by explicitly recognising that people living with rare diseases experience health inequality as a result of the rarity of their condition, including delayed diagnosis, limited clinical expertise, fragmented care and reduced access to treatments and research. This issue is increasingly well evidenced and widely acknowledged across the rare disease community. Two resources to consider are: Seeing Is Believing: Invisibility Exacerbates Inequality for Patients Living with Rare Disease - European Medical Journal; SHCA publishes a new report on rare disease health inequalities - SHCA. The significance of this issue is reflected in this year's Rare Disease Day theme, which focuses on equality in recognition of the persistent health inequalities faced by people living with rare diseases.

6	Question 5	A key challenge in implementation is the lack of clarity on how progress will be monitored. It is unclear whether there will be a national monitoring or local reporting mechanism, who will be responsible for oversight and how frequently progress will be reviewed. Monitoring will need to reflect the specific needs of the various rare diseases, and it is important to note that it will differ from previous quality standards of individual disease areas. Importantly, monitoring must include formal input from patients and families alongside other stakeholders, recognising "softer" outcomes and qualitative improvements. As with all monitoring, this should not be a purely technical box-ticking exercise but a mechanism to provide feedback and effect real positive change. Another major challenge relates to the communication and education of healthcare professionals. There is no single point of entry into the rare disease pathway, and routes to specialised care can begin anywhere in the system, including primary care, emergency settings or multiple secondary specialities, depending on presentation. Application of these quality standards needs to be universally applied, with all healthcare professionals being supported to meet their ambition. Without a coordinated approach to education, supported by national direction and local practical resources, implementation risks being inconsistent and dependent on individual awareness rather than system-wide change.
7	Statement 1	The wording of this statement is unclear and does not read easily as written, suggesting that a word or clarification may be missing. While the definition of terms later in the document explains what is meant by “first definitive treatment” and “other clock stop”, this is a public-facing quality standard and the statement itself should be understandable without needing to cross-reference definitions. There is also a lack of clarity about how national maximum waiting times should be applied in practice, given that many rare diseases do not have established pathways, guidelines or clearly defined diagnostic routes. Without this, it is difficult to understand how services are expected to operationalise the statement consistently. We remain concerned about the use of the term “definitive treatment” in the context of rare disease. Misdiagnosis is common, and premature treatment based on an incorrect or incomplete diagnosis can cause harm. For example, in conditions such as tuberous sclerosis complex, focusing on the most obvious presenting symptom and treating it as a standalone diagnosis can delay correct diagnosis and lead to inappropriate management. In addition, not all rare diseases are genetic, and whole genome sequencing cannot be relied upon as a definitive diagnostic test in all cases.

		While we agree with the rationale, the statement would benefit from greater emphasis on diagnostic accuracy, review and caution against early closure, rather than implying a linear progression to “definitive” treatment that may not reflect real-world rare disease pathways.
8	Statement 2	The statement would be strengthened by explicitly recognising that a care pathway to support future diagnosis could, where appropriate, include research. Making this explicit would better reflect the realities of rare disease care and help ensure that people are consistently informed about, and supported to access, appropriate research opportunities as part of their ongoing diagnostic journey.
9	Statement 3	We agree with the intent of this statement but have concerns about reliance on PIF TICK–accredited sources alone. For people living with ultra-rare conditions, relevant patient organisations are often small and reflect the size of the patient population. The PIF TICK process can be cost-prohibitive and time-consuming for these organisations, meaning they may not be accredited despite providing high-quality, trusted and highly specialised support. Limiting signposting to PIF TICK–accredited organisations risks excluding people with ultra-rare conditions from accessing appropriate information and support. The statement would benefit from a more flexible approach that recognises and values credible, patient-led organisations beyond formal accreditation schemes.
10	Statement 4	We agree fully with this statement and have no additional comments.
11	Statement 5	We strongly support this statement. Its impact would be further strengthened if the role of the named healthcare professional explicitly included liaison with social services and other relevant agencies, for example, around aids, adaptations, practical support and education, health and care plans (EHCPs). This would help reduce the burden on individuals and families of repeatedly retelling their story and improve coordination across systems.
12	Statement 6	The statement would benefit from clarification on whether holistic care is intended to cover only the person living with a rare disease or also their immediate family. Many rare genetic conditions present in childhood, meaning parents or carers are responsible for decision-making and often experience significant psychological and emotional impact themselves. It should be made explicit that assessment and support extend to families and carers, where appropriate, to reflect the realities of rare disease care and avoid unmet support needs.
13	Statement 7	We agree with this statement. However, we are concerned that the rapid pace of change in the availability of treatments and interventions for ultra-rare conditions means that awareness is not consistent across the healthcare system, an issue that has been reflected to us directly by healthcare professionals. Greater attention is needed on how information about newly available or recommended treatments is

		communicated clearly and consistently across all parts of the system. While monitoring uptake is important, further clarity is needed on how often this monitoring will take place, where results will be reported, the level at which data will be presented, and who will be responsible for acting on the findings.
14	Statement 8	The use of the term “care providers” in the rationale is potentially confusing and would benefit from clarification, as it could be interpreted as referring to family members or care workers rather than healthcare providers. Assuming the intention is to refer to healthcare providers, this should be stated explicitly for clarity. Otherwise, we agree with this statement.

Insert more rows as needed

Checklist for submitting comments

- Use this form and submit it as a Word document (not a PDF).
- Complete the disclosure about links with, or funding from, the tobacco industry.
- Combine all comments from your organisation into 1 response. We cannot accept more than 1 response from each organisation.
- Do not paste other tables into this table – type directly into the table.
- **Clearly mark any confidential information or other material that you do not wish to be made public. Also, ensure you state in your email to NICE that your submission includes confidential comments.**
- Do not include medical information about yourself or another person from which you or the person could be identified.
- Spell out any abbreviations you use

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Comments received from registered stakeholders and respondents during our stakeholder engagements are published in the interests of openness and transparency, and to promote understanding of how recommendations are developed. The comments are published as a record of the comments we received, and are not endorsed by NICE, its officers or advisory Committees.