

**UK Strategy  
for Rare  
Diseases**

**NHS England  
Statement of  
Intent**



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**Publications Gateway Reference: 01208**

<b>Document Purpose</b>	Guidance
<b>Document Name</b>	UK Strategy for Rare Diseases - NHS England Statement of Intent
<b>Author</b>	NHS England
<b>Publication Date</b>	26 February 2014
<b>Target Audience</b>	Patients and public

**Additional Circulation List**

**Description** This document sets out how NHS England will play its part in delivering the UK Strategy for Rare Diseases in England. A more detailed plan will be built into the five year strategy, for specialised services, currently being developed by NHS England.

**Cross Reference** UK Strategy for Rare Diseases

**Superseded Docs**  
(if applicable) N/A

**Action Required** N/A

**Timing / Deadlines**  
(if applicable) N/A

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**Document Status**

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## UK STRATEGY FOR RARE DISEASES

### NHS ENGLAND STATEMENT OF INTENT

#### INTRODUCTION

The government has issued a UK Strategy for Rare Diseases, containing 51 commitments (labelled 'C') which all four countries of the UK have agreed to achieve by 2020.

The present document sets out how NHS England will play its part in delivering those commitments.

#### EMPOWERING THOSE AFFECTED BY RARE DISEASES

***C1. Strengthen the mechanisms and opportunities for meaningful and sustained patient involvement in rare disease service provision and research, recognising patient groups as key partners – including in the development of the four country plans to implement the Strategy.***

There is always more that can be done but patient involvement is a strong theme in the way that NHS England organises its business. Specific to specialised services, there is patient representation on the 75 Clinical Reference Groups that agree service policies and specifications, and also on the Rare Disease Advisory Group.

NHS England will strengthen its relationship with patient representatives by inviting all patient representatives on Clinical Reference Groups, on an annual basis, to suggest ways in which NHS England can improve and build upon patient involvement in rare disease service provision in a meaningful and sustained way.

Each Clinical Reference Group will ensure involvement and appropriate engagement with patient organisations for rare diseases that fall within their remit.

The Rare Diseases Advisory Group will be charged with ensuring communication is carried out in a structured and systematic way. Responsibilities will include: collating information, communicating responses and circulating an action plan if necessary.

***C2. Improve awareness amongst service providers and others of the effects that rare diseases can have on a person's education, family, social relationships and ability to work.***

This improved awareness can come from close involvement of patients in service planning and delivery, as noted above.

***C3. Encourage effective and timely liaison between the NHS and other public service providers, and encourage providers to consider the effects of rare diseases on people's lives when they are developing and managing services.***

Care for patients with rare diseases, whenever possible, will be provided holistically and should include consideration of the patient's and their family's non-medical needs. All agencies involved in the care of the patient should work in an integrated way to design a pathway for patients that crosses organisational boundaries.

This should also apply to services provided across geographical boundaries and NHS England will continue to work with counterparts in the UK, including the National Services Division in Scotland, the Welsh Health Specialised Services Committee in Wales and the Health and Social Care Board in Northern Ireland to ensure patients with rare conditions can access appropriate treatment across the UK.

NHS England will also work with counterparts in Europe; this has been made possible by the EU Directive on cross-border healthcare which grants a fundamental right to all European Economic Area (EEA) citizens to access healthcare services in the EEA.

The Rare Diseases Advisory Group is best suited to oversee this work by NHS England. NHS England will report to Rare Diseases Advisory Group on the progress of cross border work on an annual basis.

***C4. Make sure that patients and their families have a say in decisions about treatment and in the planning, evaluation and monitoring of services.***

As indicated above in commitment 1, patients are already at the heart of planning, monitoring and evaluating services for rare disease through their membership of the Clinical Reference Groups and Rare Diseases Advisory Group. NHS England will empower patient representatives on Clinical Reference Groups through measures outlined in commitment 1 to make sure that membership is an active, contributing membership.

***C5. Consider how to give all patients with a rare disease clear and timely information about: their condition and its development; treatment and therapy options; practical support.***

It is already best practice for clinical teams to offer this information to patients, and the question of whether the information is clear and timely is routinely monitored in patient experience surveys.

All NHS England service specifications will be available on the NHS England website and will link to other reliable sources of information and any details of relevant patient organisations. Provider Trusts will be encouraged to provide information about their specialised services on their own websites.

***C6. Improve access for patients (or where appropriate their parents or guardians) to their personal data.***

We plan to improve access for patients to their own data in two ways. Firstly, to encourage the good practice of copying to patients all clinic letters. Secondly, by encouraging the use (subject to proper procurement) of systems such as MySpace and Patient Knows Best which allow patients to access their own data.

***C7. Support patients to register on databases, where these exist.***

Public Health England have indicated an intent to develop a national rare disease register. NHS England will play its part in helping to ensure data flows into that register.

Information regarding registries will be achieved through commitment 5.

***C8. Help patients to contribute to research and other activity related to rare diseases.***

NHS England will work with other key organisations, including the Department of Health and through the UK Rare Diseases Stakeholder Forum, to deliver this commitment.

NHS England will also contribute through its requirement that expert centres are research active (see commitment 24).

## **IDENTIFYING & PREVENTING RARE DISEASES**

***C9. Continue to work with the UK National Screening Committee to ensure that the potential role of screening in achieving earlier diagnosis is appropriately considered in the assessment of all potential new national screening programmes and proposed extensions to existing programmes.***

The UK National Screening Committee has primary responsibility for this commitment. NHS England will contribute actively as required.

***C10. Initiate action to ensure carrier testing approved by the appropriate commissioning bodies, where the associated molecular tests are evaluated and recommended by UKGTN, is accessible for at risk relatives.***

The recommendations of the UK Genetic Testing Network will be considered by NHS England and subject to available resources, incorporated into services specifications.

## **DIAGNOSIS & EARLY INTERVENTION**

***C11. Work to achieve reduced times for diagnosis of rare diseases, whilst acknowledging that more needs to be done to ensure that undiagnosed patients have appropriate access to coordinated care to help disabled children who are thought to have a genetic syndrome or condition that science has not yet identified.***

Implementation of this commitment will be achieved in part through the mechanisms set out in commitment 12 and 23.

We will also develop a working group to identify the problems associated with the diagnosis of rare diseases. The working group will report back, within 12 months, on the ways in which NHS England can improve diagnosis and early intervention. This working group will initially be a sub group within the Rare Diseases Advisory Group with the intention of broader membership.

***C12. Work with the NHS and clinicians to establish appropriate diagnostic pathways which are accessible to, and understood by, professionals and patients, by:***

- ***establishing clear, easily accessible and effective pathways between primary care, secondary care, regional centres and specialist clinical centres, as appropriate***
- ***putting protocols in place to identify patients with no diagnosis, ensuring that a lack of diagnosis does not create a barrier to treatment***
- ***drawing on patients' ability to help inform decisions about referral and diagnosis***
- ***creating effective clinical networks to support this process***
- ***making high quality diagnostic tests accessible through common, clinically agreed systems or pathway***
- ***embedding appropriate information in national data systems including measuring equity of access to molecular tests to maintain UKGTN diagnostic studies.***

This commitment will be achieved primarily through the Rare Disease Annex for all service specifications, which all Clinical Reference Groups must consider when commissioning specialised services. The Rare Disease Annex will incorporate all points listed above. Effective clinical networks will also be delivered through commitment 26 and 27.

The Rare Diseases Advisory Group has been tasked to oversee the effectiveness of the Rare Disease Annex. NHS England will provide the necessary information to allow the Rare Diseases Advisory Group to carry out monitoring duties.

***C13. Ensure that there are appropriate procedures for evaluating the costs and benefits of treatments for patients.***

NHS England will collaborate closely with the National Institute for Health and Care Excellence (NICE) in the formal appraisal of technologies, including the newly established evaluations committee for highly specialised technologies. NHS England's internal procedures for evaluating the costs and benefits of treatment will, where appropriate, take account of the specific needs of patients with rare disease. These evaluations will be publicly available for scrutiny.

***C14. Where appropriate, support the availability of computerised prompts to help GPs diagnose a rare disease when a rare disease has not previously been considered***

Expert systems show great promise as a tool to help early recognition of rare disease. This technology is however not yet fully developed. NHS England will support piloting and evaluation where appropriate.

***C15. Improve education and awareness of rare diseases across the healthcare professions, including:***

- ***involving patients in the development of training programmes***
- ***encouraging medical, nursing and associated health professionals to get hands-on experience in specialist clinics***
- ***ensuring awareness of methods and clinical techniques used in differential diagnosis***

*NHS England will provide appropriate support to those leading on the delivery of this commitment*

***C16. Monitor the development of ICD-11 in preparation for its adoption***

The strategic intent of NHS England is to adopt SNOMED CT as its standard coding system. SNOMED CT is interoperable with ICD 10, and will be with ICD 11 when developed.

***C17. Work with colleagues in Europe on the development of the European Orphanet coding system and considering the adoption of Orphanet coding and nomenclature***

NHS England will deliver this commitment and we are currently investigating appropriate methods to do so.

***C18. Standardise data collection, building upon existing NHS data standards, and develop standards where they do not exist, increasing the reliability of information for use in providing or commissioning care***

NHS England is currently in discussion with relevant agencies in order to establish responsibility for this commitment.

***C19. Explore options to improve the link between existing patient data and electronic health records***

NHS England is currently in discussion with relevant agencies in order to establish responsibility for this commitment.

***C20. Assess the potential for rare disease databases where they do not exist***

As noted in commitment 7, Public Health England have indicated an intent to develop a national rare disease register which will cover every patient in England with a rare disease. NHS England will play its part in helping to ensure data flows into that register.

***C21. Agree international standards, building on existing NHS standards***

NHS England is represented on the international body which governs health data standards, the *International Health Terminology Standards Development Organisation*. We will build on this platform to deliver this commitment.

***C22. Support international links to UK databases and build on the work of current funded programmes that aim to link rare disease research internationally***

NHS England will deliver this commitment and we are currently investigating appropriate methods to do so.

## **COORDINATION OF CARE**

***C23. Continue to develop service specifications for rare diseases. This will include country specific care pathways and a 'generic' care pathway that sets out best practice that can be applied to all patients with rare diseases in the UK (particularly where there are no disease specific pathways). The generic care pathway will include:***

- ***an appropriate care plan for all patients with a rare disease***
- ***clearly stated principles around the standards of care which patients with a rare disease can expect, including patients with no diagnosis***

- ***the development of seamless pathways for transition, from childhood to adolescence, and on to adulthood and older age***
- ***access criteria and measures of quality and outcomes***

As noted in commitments 3 and 12 NHS England will develop a Rare Disease Annex that will apply to all service specifications and will incorporate the requirements listed above. It will also incorporate an assurance that every patient will be made aware of who is responsible for providing their care. Provisions for patients with undiagnosed conditions will also be made.

***C24. Agree that specialist clinical centres should as a minimum standard:***

- ***have a sufficient caseload to build recognised expertise***
- ***where possible, not depend on a single clinician***
- ***coordinate care***
- ***arrange for coordinated transition from children's to adults' services***
- ***involve people with rare conditions, and their families and carers***
- ***support research activity***
- ***ensure their expertise is available to families and their healthcare teams***

*NHS England will reflect these requirements in the Rare Disease Annex which will apply to centres providing specialist care for rare diseases.*

***C25. Ensure that the relationship between the specialist clinical centres and science and research is explained and understood and put into practice by: practitioners delivering local health and social care; the research community; industry; academia***

NHS England will work with other key organisations, including the Department of Health and through the UK Rare Diseases Stakeholder Forum, to deliver this commitment.

We will encourage specialist centres to make appropriate links to research networks and centres.

***C26. Set out clearly the connections to and communications with specialist clinical centres in molecular diagnostics and other forms of diagnostic support***

NHS England will work with other key organisations, including the Department of Health and through the UK Rare Diseases Stakeholder Forum, to deliver this commitment.

***C27. Ensure that specialist clinical centres are as concerned with research as with health and social care support, and that they develop networks that provide professional to- professional dialogue and collaboration across a wide range of experts, including internationally (especially for those conditions that are ultrarare)***

NHS England will work with other key organisations, including the Department of Health and through the UK Rare Diseases Stakeholder Forum, to deliver this commitment.

***C28. Work with international partners wherever possible and develop UK-wide criteria for centres to become part of an expert reference network to increase the flow of information between patients and professionals in a range of disciplines***

The European Commission is developing a system for Europe-wide networks of expert reference centres, and is likely to develop, for each network, disease specific criteria. Within England the general criteria for centres will be as set out in commitment 23 above, and developed more specifically in the Rare Disease Annex.

***C29. Improve systems to record genetic and other relevant information accurately to record the incidence and prevalence of disease and support service planning and international planning***

This benefit will flow from the establishment of a national register for rare disease – see commitment (C) 7 above.

***C30. Identify how they can change systems to hold information about rare diseases, including information about the uptake of treatments***

The main limitation at present is the lack of a good coding system for rare disease. SNOMED CT overcomes this limitation – see commitment 16.

## **THE ROLE OF RESEARCH**

***C31. Look at how the 4 UK countries develop, change or expand information systems to capture, connect and analyse data about clinical and social care pathways***

NHS England will work with other key organisations, including the Department of Health and through the UK Rare Diseases Stakeholder Forum, to deliver this commitment.

***C32. Work together to identify a selection of the rare diseases most suited to the development of best-care pathways and propose other rare diseases for possible pathway development, taking on board the needs of patients and carers and the challenges faced during delivery of the first set of pathways***

NHS England will carry out a priority setting exercise to identify the most suitable pathway development; this will involve patient groups.

***C33. Examine how they can encourage service providers to involve patients in research and to ensure appropriate funding for excess treatment costs for research in rare diseases***

*The NHS Standard Contract makes reference to excess treatment costs and we are producing a research resource kit for commissioners.*

*NHS England will also support existing projects that encourage the increased involvement of patients in research design and participation in clinical trials.*

***C34. Make better use of online applications to give patients information about their condition so that they can develop a personalised care path plan with their clinical and social care team***

NHS England will deliver this commitment and we are currently investigating appropriate methods to do so.

***C35. Use portals to connect patients and relatives to enhance research participation and, where appropriate, promote self-enrolment to approved research studies with online consenting, self-reporting and use of social media***

*NHS England will work with other key organisations, including the Department of Health and through the UK Rare Diseases Stakeholder Forum, to deliver this commitment.*

***C36. Encourage patient groups to get involved with regulatory bodies***

*NHS England will provide appropriate support to those leading on the delivery of this commitment.*

***C37. Help patient organisations and community engagement events develop more formal partnerships with the NHS research-active organisations***

*NHS England will work with other key organisations, including the Department of Health and through the UK Rare Diseases Stakeholder Forum, to deliver this commitment.*

***C38. Explore the feasibility of the UK Clinical Trials Gateway including experimental medicine trials for rare diseases to provide information for patients and their families about research trials***

*NHS England will provide appropriate support to those leading on the delivery of this commitment.*

**C39. Work with the research community, regulators, providers of NHS services and research funders to develop risk-proportional permission systems**

*NHS England will provide appropriate support to those leading on the delivery of this commitment.*

**C40. Encourage researchers to use current guidance to produce generic participant information leaflets and consent forms and participate in future guidance reviews.**

*NHS England will provide appropriate support to those leading on the delivery of this commitment.*

**C41. Promote good practice and the use of systems which facilitate a consistent and streamlined process to local NHS permissions of publically, charitably and commercially funded research with an aim to reduce timescales.**

*NHS England will provide appropriate support to those leading on the delivery of this commitment.*

**C42. Begin and complete next generation sequencing (NGS) demonstration projects to: evaluate their usefulness, acceptability and cost-effectiveness; develop effective health economic assessments (for example through Health Technology Assessments) and similar initiatives**

*NHS England will provide appropriate support to those leading on the delivery of this commitment.*

**C43. Evaluate different NGS platform configurations, for example:**

- **NGS for clinical condition-specific sets of genes (such as 100–200 of the**
- **22,000 genes**
- **whole exome sequencing (2% of the entire genome)**
- **whole genome sequencing**

*NHS England will work with other key organisations, including the Department of Health and through the UK Rare Diseases Stakeholder Forum, to deliver this commitment.*

**C44. Support the introduction of NGS into mainstream NHS diagnostic pathways, underpinned by appropriate clinical bioinformatics, including clinical bioinformatics hubs supported by high performance computing centres, where appropriate**

*NHS England will provide appropriate support to those leading on the delivery of this commitment.*

**C45. Ensure that training and education are available to the NHS workforce, highlighting the importance of NGS to all aspects of rare disease care, including support for evidence based local counselling for patients and their relatives who receive NGS results**

*NHS England will provide appropriate support to those leading on the delivery of this commitment.*

**C46. Work with industry to set priorities and determine how best to support research into rare diseases and promote research collaboration**

*NHS England will provide appropriate support to those leading on the delivery of this commitment.*

**C47. Support initiatives to facilitate engagement between patients, clinical care teams, researchers and industry wherever practical**

*NHS England will work with other key organisations, including the Department of Health and through the UK Rare Diseases Stakeholder Forum, to deliver this commitment.*

**C48. Set out the benefits of collaboration (besides producing specific treatments) for all Stakeholders**

To deliver this commitment NHS England will work with key stakeholders, including: Industry; patients and relatives; main funding providers; healthcare commissioners; and NHS hospitals and specialist care units to develop a compact.

**C49. Continue to build a cohesive infrastructure for implementation and coordination of rare disease research in the NHS**

*NHS England will provide appropriate support to those leading on the delivery of this commitment.*

**C50. Encourage major research funders to use current structures to coordinate strategic funding initiatives in rare diseases**

*NHS England will provide appropriate support to those leading on the delivery of this commitment.*

**C51. Improve engagement between key stakeholders, including:**

- a) patients and relatives**
- b) main funding providers**
- c) healthcare commissioners**

*NHS England will work with other key organisations, including the Department of Health and through the UK Rare Diseases Stakeholder Forum, to deliver this commitment.*