



# **Implementation Plan for the UK Strategy for Rare Diseases**

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# **Implementation Plan for the UK Strategy for Rare Diseases**

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## Executive summary

NHS England, through its role in the direct commissioning of specialised services, is ideally placed to improve the care of patients who have rare diseases.

This is the Implementation Plan setting out NHS England's delivery contribution to the UK Strategy for Rare Diseases.

The Plan sets out NHS England's proposed actions against all of the commitments in the Strategy for which it has a lead responsibility. In particular, the Plan aims to address the following three objectives:

- Facilitating earlier diagnosis and intervention.
- Improving care coordination.
- Promoting research.

Of particular note are the following key actions:

- The continuing progress of the 100,000 Genomes Project and the concurrent development of a genomic testing strategy that will underpin the development of a new genomic medicine services for the NHS
- The development of a set of criteria that will allow NHS England to hold providers to account for the way in which they treat patients with rare diseases via a rare disease 'insert' to the standard NHS Contract
- The development of Rare Disease Collaborative Networks. These will be groups of providers who have a demonstrable research-active interest in a rare/very rare disease, with the aim of improving patient outcomes

This Plan should be read in conjunction with the Department of Health's overall plan, which sets out the broader set of actions being taken by other parts of the health and care system against the commitments in the Strategy.

We are very grateful to the stakeholders who have worked with NHS England to develop this Plan. In particular, we would like to thank Genetic Alliance UK and the Specialised Healthcare Alliance for listening to our ideas and reviewing earlier drafts of the Plan. The Plan has very much benefited from these contributions; for example, there was a clear message that we needed to be able to measure progress and this is reflected in the dashboard.

## Equality and Health Inequalities Statement

Promoting equality and addressing health inequalities are at the heart of NHS England's values. Throughout the development of the policies and processes cited in this document, we have:

- Given due regard to the need to eliminate discrimination, harassment and victimisation, to advance equality of opportunity, and to foster good relations between people who share a relevant protected characteristic (as cited under the Equality Act 2010) and those who do not share it; and
- Given regard to the need to reduce inequalities between patients in access to, and outcomes from healthcare services and to ensure services are provided in an integrated way where this might reduce health inequalities.

### 1. Background

The UK Strategy for Rare Diseases (the Strategy) was published by government in November 2013 with the objective of 'setting out a shared vision for the UK to improve the lives of those with rare diseases'.<sup>1</sup>

In the document 'Next Steps on the NHS Five Year Forward View',<sup>2</sup> published on 31 March 2017, NHS England confirmed that it would 'publish an Implementation Plan setting out its delivery contribution to the UK Strategy for Rare Diseases.

There are between 5,000 and 8,000 rare diseases that affect the lives of around three million of the UK population. These can present in childhood or adulthood and can be both genetic and non-genetic in origin. The Strategy defines a rare disease as 'a life-threatening or chronically debilitating disease that affects five people or fewer in 10,000 and requires special, combined efforts to enable patients to be treated effectively'.<sup>1</sup> In England, this equates to a disease that affects 25,000 or fewer people.

This document sets out our delivery contributions to 2020 and the document will be refreshed over time. As many of our existing work programmes impact on patients with rare diseases, this document is intended to set out those activities that are specifically associated with implementing the Strategy.

### 2. Statutory framework and governance

The National Health Service Act 2006 as amended by the Health and Social Care Act 2012 sets out that NHS England has statutory responsibility for commissioning a list of 'services for rare and very rare conditions'. This list of services is amended from time to time and the latest list was published in July 2017.<sup>3</sup>

Whilst we have responsibility for commissioning these specialised services, the majority of care for patients with rare diseases is delivered – entirely appropriately – locally, with many elements commissioned by Clinical Commissioning Groups.

The list of 150 specialised services that we commission includes some services for patients with rare diseases but also many interventional services, such as specialist surgery. Within the 150 specialised services, there are 70 *highly* specialised services.<sup>4</sup> These are provided to a smaller number of patients compared to specialised services; usually no more than 500 patients per year. For this reason, they are delivered nationally through a very small number of expert centres. As with specialised services, the highly specialised services portfolio includes some services for patients with very rare diseases and some interventional services such as highly specialist surgery.

The Rare Diseases Advisory Group (RDAG)<sup>5</sup> makes recommendations to NHS England and the devolved administrations of Scotland, Wales and Northern Ireland on developing and implementing the strategy for rare diseases and highly specialised services. The membership of RDAG is broad and includes representatives from Royal Colleges, commissioners, patient and public voice representation and professionals such as a geneticist and an ethicist.

The terms of reference for RDAG include as a duty: *‘To make recommendations to NHS England and the devolved administrations on how the UK Strategy for Rare Diseases should be implemented.’*

The Rare Disease Policy Board (RDPB)<sup>6</sup> is a UK-wide committee, managed by the Department of Health, with responsibility for facilitating the coordination of policy development and meeting the commitments set out in the Strategy.

In summary, the role of the RDPB is to monitor *what* is being implemented in respect of the Strategy and the role of RDAG is to make recommendations to NHS England and the devolved administrations on *how* the Strategy is implemented. We are committed to reporting progress against the Strategy to the RDPB through our membership of that committee. NHS England will also work within any arrangements that are established to facilitate progress against the implementation of the Strategy specifically in England.

### 3. Implementing the commitments in the Strategy

The Strategy is divided into five areas:

- Empowering those affected by rare diseases
- Identifying and preventing rare diseases
- Diagnosis and early intervention
- Coordination of care
- The role of research

There are 51 commitments in the Strategy, for which NHS England has a lead (for England) on 31 (one commitment has been completed).

For each commitment where NHS England is a lead organisation, there is a performance measure in the dashboard (see Appendix A). This is so that we can demonstrate that we are making progress towards implementing that commitment.

In this Implementation Plan, NHS England's proposed actions are divided into two parts:

- Three key objectives (against which the majority of those commitments for which NHS England has delivery responsibility map); and
- A list of 'enablers' that describe how we are meeting the commitments in the Strategy through our 'business as usual' processes.

## 4. The three key objectives

The three key objectives in this Implementation Plan are:

- Facilitating earlier diagnosis and intervention
- Improving care coordination
- Promoting research

Whilst these objectives broadly map to the five areas in the Strategy, they reflect our specific delivery contributions to the Strategy.

## 5. Objective 1 – Facilitating earlier diagnosis and intervention

Facilitating earlier diagnosis and intervention will be fundamental to meeting the commitments in the Strategy. On average, patients with a rare disease will consult with five doctors; receive three misdiagnoses; and wait four years before receiving their definitive diagnosis.<sup>7</sup> Delays in diagnosis mean that opportunities for timely interventions can be missed; conversely, patients may be given inappropriate or harmful treatments if they have been given the wrong diagnosis. Timely diagnosis also allows for the making of reproductive choices by couples when a disease is known to be heritable.

Getting a correct diagnosis reduces the time that patients and families have to spend searching and visiting a multiplicity of healthcare professionals. Improving diagnosis means patients and families can be signposted to services that are expert in the care of their disease. Other agencies such as education and social services can be reluctant to commit to specific interventions without a diagnosis.

In her annual report published on 4 July 2017, the Chief Medical Officer called on *“clinical staff, managers and the Government to work together to make wider use of revolutionary genetics techniques in the battle to improve cancer survival rates and identify rare diseases faster so patients can get the right care at the earliest opportunity”*.<sup>8</sup>

The 100,000 Genomes Project,<sup>9</sup> which began in 2012, is not only on target to sequence 100,000 genomes through the 13 Genomic Medicine Centres, but is also contributing towards building the infrastructure for routine commissioned genomic medicine in the NHS.

The core actions we intend to take to implement this objective are as follows:

- We will continue to **develop and implement our genomic testing strategy** for the NHS, including determining which tests will be recommended for which disease from single gene tests to multiplex panels and whole genome sequencing. This will be undertaken through the Rare Disease Transition Working Group. [2017/18 through 2019/20] [C10, C43, C44]
- We will continue to identify a list of specific interventions that can be taken to **reduce delays in diagnosis**, for example, identifying additional genetic tests for rare diseases that can be added to existing panel (genetic) tests. We will need to understand the clinical and cost effectiveness of these interventions within the context of wider pressures on NHS budgets. The pace and sequence of their implementation will depend on the extent to which they describe a pathway in which unnecessary steps are removed, or timeliness is improved or costs are reduced. [2018 through 2020] [C11]
- We will talk to the Royal College of General Practitioners and the Nursing & Midwifery Council about opportunities for GPs and health visitors, for example, to **facilitate earlier diagnosis** of rare diseases. We would pilot any proposals to ensure their effectiveness. [2018/19] [C14]
- We will **re-procure the genomic infrastructure** across the NHS. [2017 through 2019] [C26] In particular, in implementing the service specification, we will ensure that there is active patient and public engagement in identifying ways of reducing inequity of access. [2017/18] [C1]

In addition:

- We will take forward the recommendation in *Developing a consensus on data sharing to support NHS clinical genetics and genomics services*<sup>10</sup> to develop a privacy assessment to examine suitable arrangements for the sharing of data within the NHS where genomic testing could be used as part of routine clinical care. [2018 through 2020] [C6]
- We will continue to support the NHS Genomic Medicine Centres that have been established through the 100,000 Genomes Project to:
  - Establish new genomic medicine pathways
  - Involve more hospitals in identifying rare diseases
  - Secure greater involvement of other clinical specialties in identifying and characterising rare diseases
  - Design and test optimal pathway arrangements for patients with rare diseases. [2018 through 2019] [C10, C44]
- We will continue to contribute to the Diagnostic Odyssey Task & Finish Group (a sub-committee of the RDPB). [2017/18] [C11]
- We will support the Academy of Medical Royal Colleges in developing a Genomics Champions' Network to disseminate information about the value of

genomic testing to mainstream clinicians and when genomic testing may be useful in the patient pathway. [2018 through 2020] [C11]

- We will continue to monitor the development of ICD-11 and any derivative products, particularly in relation to SNOMED CT. [2018 through 2020] [C16]
- We will continue to contribute towards the work to harmonise Orphanet and SNOMED, noting that a more global option may be to link OMIM and SNOMED. [2018 through 2020] [C17]
- We will continue to standardise data collection, including through the genomic testing strategy. [2018 through 2020] [C18]
- We will continue to adopt SNOMED as our preferred system for clinical terminology. [2018 through 2020] [C21]
- Initially through the involvement of the West Midlands Academic Health Science Network (AHSN) in the genomics agenda, other AHSNs may become involved in the rare disease agenda. [2018/19] [C25]
- We will continue to support the accurate characterisation of patients through Human Phenotype Ontology terms as being developed through the 100,000 Genomes Project. [2018 through 2020] [C44]

Stakeholders can help us to implement the Plan by:

- Letting us know about additional specific interventions that can be taken to reduce delays in diagnosis

## 6. Objective 2 – Improving care coordination

Improving care coordination will be an important element of improving care for people with rare diseases. In their report *Rare Disease Care Coordination: Delivering Value, Improving Services*, 2013,<sup>11</sup> Rare Disease UK found strong evidence that care coordinators represent good value for money for service providers, for example, by: saving consultants' and GPs' time; helping to prevent unplanned hospital admissions; and reducing the length of hospital stays.

They also found that having a professional in post who can fulfil the care coordinator role helps to improve quality of care and patients' experience of care, for example, by: helping patients receive timely access to the specialist knowledge and care they require; meeting patients' information needs; providing emotional and practical support to patients and their families; and providing a continuing point of contact. Improving coordination in this way will also support the empowerment of patients – a key element of the Strategy.

In their report *The Hidden Costs of Rare Diseases: A Feasibility Study* 2016,<sup>12</sup> Genetic Alliance UK found that poorly coordinated care is a major issue for patients and families affected by rare conditions, with 1 in 3 patients attending three or more

clinics for their condition and more than 8 in 10 patients not having a care coordinator or advisor.

There are a number of *Regulation 28 Reports To Prevent Future Deaths* (for example, Arrowsmith 2015 and Ashton 2014<sup>13</sup>) where a provider did not take account of information about a patient's rare disease and where this unfamiliarity may have contributed towards the patient's death.

In their report '*Patient experiences of transition between care providers, 2014*',<sup>14</sup> Rare Disease UK highlighted a number of aspects of transition that were unsatisfactory including: patients and families feeling disconnected from the transition process; age-appropriate services not always being available to patients; medical professionals receiving insufficient training in adolescent care and medicine; research studies not being compatible with the transition process; transition coming as a shock to patients and being too sudden; parents feeling anxious and unsupported during transition; patients' individual circumstances not always being considered during transition.

The core actions we intend to take to implement this objective are as follows:

- We will develop [2017/18] and implement [2018/19] a **rare disease 'insert'**. This will be a set of criteria that will sit alongside those NHS England service specifications (see enablers) for services that treat patients with rare diseases. These criteria will allow us to hold providers to account for the way in which they treat patients with rare diseases. There will be up to three criteria in the insert (depending on the nature of the service):
  - That the provider must ensure that there is a person responsible for **coordinating the care** of any patient with a rare disease [C2, 3, 24]
  - That the provider must give every patient with a rare disease an '**alert card**'(including information about: the patient's rare disease; any particular aspects of the treatment of that rare disease that need to be taken into account in providing care to that patient; and details of how to contact an individual expert in that patient's care) [C3]
  - That the provider must ensure that every paediatric patient with a rare disease has an **active transition to an appropriate adult service**, even if that adult service is not the commissioning responsibility of NHS England [C24]
- We will explore with the Royal College of Emergency Medicine how alert cards could be clinically **recognised in A&E departments**. [2018/19] [C3]
- We will work with providers on **new models** for the treatment of rare diseases, for example, the rare disease centres being established in Birmingham and London. [2018/19] [C24]

In addition:

- We will evaluate the impact of the rare disease insert and consider whether it has a wider potential for other NHS contracts. [2018/19] [C2, 3, 24]

- We will explore with NHS Digital the benefits and feasibility of inserting a ‘red flag’ onto the summary care record of any patient with a rare disease whose life would be at risk if they were treated incorrectly. [2017/18] [C3]
- We will reform outpatient tariffs to support new care models. We will pilot any proposals to evaluate their effectiveness. [2018/19] [C24]
- We will use the newly developed ‘aspirant market entrant process’ for highly specialised services; this gives providers the opportunity to demonstrate a case for there being additional/replacement providers of services, for example, because there is inequitable geographical access to a service. [2018 through 2020] [C24]
- We will explore options for using SNOMED and point of care recording to minimise manual data entries, thereby: making cost efficiencies; creating true secondary use of primary input data; and sharing key information to facilitate care coordination. [2018/19] [C30]

Stakeholders can help us to implement the Plan by:

- Giving us examples of alert cards and sharing their experiences of attending unfamiliar providers
- Helping us to explore, with the Royal College of Emergency Medicine, how alert cards will be clinically recognised in A&E departments

## 7. Objective 3 – Promoting research

Finally, we need to continue to play an important role in promoting research. We want to enable professional to professional dialogue within the UK and provide a recognised focus for international collaboration.

We want patients to be able to make an informed choice about those providers that are likely to be most expert in the care of their rare disease. Providers who are research active in a particular rare disease tend to be likely to be able to deliver better and more coordinated care to patients and with improved outcomes.

In November 2017, the NHS England Board considered a paper that was developed jointly with the National Institute for Health Research entitled ‘Twelve actions to support and apply research in the NHS’.<sup>15</sup> A number of the actions have relevance to patients with rare diseases, in particular, to:

- Manage excess treatment costs better
- Eliminate delays in confirming multi-site trials
- Develop the NHS genomic medicine service

- Use NHS England's specialised commissioning and commercial medicines clout, combined with NICE appraisals, to drive faster uptake of affordable, high impact innovation

The core actions we intend to take to implement this objective are as follows:

- We will develop [2017/18] and implement [2018/19] a process for recognising **Rare Disease Collaborative Networks** (RDCNs), with NHS England defining a RDRN as a 'recognised network of member providers, each of which has a demonstrable research-active interest in a rare/very rare disease, the aim of the network being to improve patient outcomes'. NHS England defines a Rare Disease Collaborative Centre as 'a provider that has been recognised as having a demonstrable research-active interest in a rare/very rare disease and who works with other recognised providers in a network to improve patient outcomes.' [C8, 27, 33, 47, 51]
- In line with the paper considered at the NHS England Board in November 2017, we are proposing three changes to manage excess treatment costs (ETCs) better, the first two of which will particularly impact on patients with rare diseases:
  - We will develop a more rapid, standardised process for specialised commissioning ETCs
  - We will set a minimum threshold under which excess treatment costs must be absorbed by providers.
  - We will utilise the 15 NIHR Local Clinical Research Networks (LCRNs) to help manage the process for ETCs on behalf of their local CCGs. [C33]

In addition:

- We will explore the use of the Genomics England National Data Set with a view to using this alongside other datasets to enhance patient care. [2018/19] [C8]
- We will continue to support the implementation of European Reference Networks. [2018 through 2020] [C22, 27, 28]
- We will explore the development of apps for patients with rare diseases through the NHS Digital Apps Library; these may be apps that are specific to individual rare diseases or generics apps for example for parents or children who have a rare disease. It may be appropriate that some apps are designed for those clinicians who do not customarily treat patients with rare diseases and need information to treat them appropriately. [2018/19] [C34]
- We will continue to support, in partnership with Genomics England, the UK Genomic Knowledge Base that has been developed through the 100,000 Genomes Project and which will assist with variant classification in rare diseases. [2018 through 2020] [C44]

Stakeholders can help us to implement the Plan by:

- Giving us examples of apps for patients with rare diseases

## 8. Enablers

'Enablers' are 'business as usual' processes that contribute towards us meeting the commitments in the Strategy:

- We will continue to use the *Framework for patient and public participation in specialised commissioning*,<sup>16</sup> that sets out how 163 patient and public members are involved in our governance structures. In addition NHS England involves patients on the working groups that develop clinical commissioning policies and service specifications. As well as involving patients through these mechanisms, we have well-developed processes for both engaging patients and the public when developing these commissioning products (through the registered stakeholder structure) and for public consultation. [C1]
- We will continue to develop service specifications in line with our *Methods: National Service Specifications*<sup>17</sup> document. The template for service specifications includes sections on: 'Outcomes and applicable quality standards' [C4, 5, 12, 23]; care pathways [C5, 12, 23, 32]; evidence base [C5]; and service standards [C23].
- We will continue to develop clinical commissioning policies in line with our *Methods: National Clinical Policies*<sup>18</sup> document. The policy proposition template includes sections on: evidence base [C5]; the patient pathway [C5, 12]. Each policy proposition also includes a plain language summary of the policy [C5].
- We and NICE will continue to evaluate the costs and benefits of treatments for patients in line with published processes, such as the *NHS England Specialised Commissioning Service Development Policy*<sup>19</sup>. [C13]

We will also continue to use the *NHS Standard Contract 2017/18-2018/19*<sup>20</sup> to hold providers to account on:

- Liaison between NHS providers and other public service providers
- Patients and their families having a say in the planning, evaluation and monitoring of services
- Provision of clear and timely information
- Provision of information to patients
- Patient involvement in research
- Provision of clinical networks
- Provision of an information schedule that sets out standards for data flows

## **9. Demonstrating progress towards implementing the commitments in the Strategy**

We will monitor the impact of the Implementation Plan by:

- Reviewing progress against the commitment-specific performance measures set out in the dashboard (see Appendix A).
- Meeting with stakeholders to report progress.
- Reporting progress to the Rare Diseases Policy Board and to any other group that is established to review progress against the implementation of the Strategy specifically in England.
- Establishing programme management arrangements within NHS England.
- Exploring whether it would be appropriate to establish a rare disease stakeholder group as is the case with NHS England's Clinical Reference Groups.
- Exploring whether it would be appropriate to establish a virtual reference group

## Appendix A – dashboard of performance measures

Measure	Commitment(s) being measured
<ul style="list-style-type: none"> <li>Number of individuals involved in the Genomics Medicine Centres patient and public involvement groups</li> <li>Examples of good practice in the Genomics Medicine Centres</li> </ul>	1
<ul style="list-style-type: none"> <li>Published service specification development process available on the NHS England website</li> <li>Number of service specifications that have been out for stakeholder engagement</li> <li>Number of service specifications that have been out for public consultation</li> <li>Number of service specifications published</li> <li>Examples of good practice in the development of service specifications</li> </ul>	1, 4, 5, 12, 23, 32 1, 4 1,4 5, 12, 23, 32 5, 12, 23, 32
<ul style="list-style-type: none"> <li>Published clinical policy development process available on the NHS England website</li> <li>Number of clinical policies that have been out for stakeholder testing</li> <li>Number of clinical policies that have been out for public consultation</li> <li>Number of clinical policies published</li> <li>Examples of good practice in the development of clinical policies</li> </ul>	1, 5, 12 1 1 5, 12 5, 12
<ul style="list-style-type: none"> <li>Number of patient and public voice individuals involved in NHS England specialised commissioning governance processes</li> <li>Examples of good practice in patient and public participation</li> </ul>	1
<ul style="list-style-type: none"> <li>% of providers compliant with rare disease insert elements</li> <li>Examples of good practice in use of the rare disease insert and how non-compliance is being addressed</li> </ul>	2, 3, 24
Progress report on the development of a privacy impact assessment to examine suitable arrangements for the sharing of data within the NHS where genomic testing could be used as part of routine clinical care	6
<ul style="list-style-type: none"> <li>Number of rare disease collaborative networks endorsed</li> <li>Number of rare disease collaborative centres endorsed</li> </ul>	8, 27, 33, 47, 51
Published Genomic Testing Strategy available on the NHS England website	10, 43, 44
Number of specific early diagnostic interventions implemented	11
Progress report on the Genomics Champions' Network	11
Published processes for the evaluation of the costs and benefits of treatments available on the NHS England and NICE websites	13
Progress report on the adoption of SNOMED	16, 17, 21
Progress report on standardisation of data collection areas	18
Number of providers endorsed as members/leads of European Reference Networks	22, 27, 28
<ul style="list-style-type: none"> <li>Number of providers engaging with NHS England through aspirant market entrant process</li> <li>Number of service reviews initiated through the aspirant market engagement process</li> </ul>	24
Reprocurement/mobilisation of the genomic infrastructure complete	26
Published process for excess treatment costs available on the NHS England website	33
Number of Whole Genome Sequencing samples collected	43, 44

## Appendix B – glossary

Term	Definition
100,000 Genomes Project	A project that will sequence 100,000 genomes from around 70,000 people, the aim of which is to create a new genomic medicine service for the NHS, transforming the way in which people receive care.
Clinical policy	A document that sets out NHS England's clinical access to a specialised treatment.
Diagnostic Odyssey Task & Finish Group	A sub-committee of the UK Rare Disease Policy Board whose objective is to establish <b>a process</b> that will measure the time travelled in the diagnostic pathway for patients with particular rare diseases.
European Reference Network	An organisation that help professionals and centres of expertise in different EU countries to share knowledge.
Excess treatment costs	The difference between total treatment costs during a clinical trial and the costs of standard NHS treatment, where total treatment costs comprise: research costs; research support costs; and NHS treatment costs (the costs that would be incurred by the NHS if the treatment under trial was to be adopted by the NHS).
Genetic Alliance UK	A national charity working to improve the lives of patients and families affected by all types of genetic conditions.
Genomics England	The organisation set up to deliver the 100,000 Genomes Project.
Genomic Medicine Centre	One of 13 providers across England that are delivering the 100,000 Genomes Project.
Human Phenotype Ontology	A standardised vocabulary of phenotypic abnormalities (i.e. those that physically expressed) and which are encountered in human disease. (The <i>genotype</i> is the set of genes in DNA that is responsible for the abnormality).
ICD-11	The International Classification of Disease 11 <sup>th</sup> revisions – the global classification system used for mortality and morbidity statistics.
Individual funding request	A request for treatment that is not normally commissioned by NHS

Term	Definition
	England.
In-year service development	Any aspect of healthcare, other than one that is the subject of an individual funding request, that NHS England funds outside the usual annual commissioning round.
NHS Digital Apps Library	A library of digital tools designed to help patients manage and improve their health.
Orphanet	A reference source of information on rare diseases located at <a href="http://www.orpha.net">www.orpha.net</a>
OMIM	Online Mendelian Inheritance in Man is a compendium of human genes and genetic phenotypes.
Rare disease	The European definition of a rare disease is a life-threatening or chronically debilitating disease that affects five people or fewer in 10,000. Rare diseases often require special efforts to co-ordinate care given by many different specialists and agencies.
Rare Disease Policy Board	A committee of the Department of Health that has responsibility for the coordination of policy development and meeting the commitments set out in the Strategy.
Rare Disease Collaborative Network/Centre	<p>A recognised network of member providers, each of which has a demonstrable research-active interest in a rare/very rare disease, the aim of the network being to improve patient outcomes.</p> <p>A provider that has been recognised as having a demonstrable research-active interest in a rare/very rare disease and who works with other recognised providers in a network to improve patient outcomes.</p>
Rare Disease Transition Working Group	An NHS England committee responsible for developing a genetic testing strategy for the NHS in England, building on the legacy of the 100,000 Genomes Project.
Rare Disease UK	The national campaign for people with rare diseases and all who support them.
Rare Diseases Advisory Group	An NHS England committee that makes recommendations to NHS England and the devolved administrations of Scotland, Wales and Northern Ireland on developing and implementing the

Term	Definition
	strategy for rare diseases and highly specialised services.
Regulation 28 Reports To Prevent Future Deaths	Reports issued by coroners to a person, organisation, local authority or government department or agency where the coroner believes that action should be taken to prevent future deaths.
Service specification	A document that sets out how NHS England requires a provider to deliver a specialised service.
SNOMED/SNOMED-CT	Systemized Nomenclature of Medicine-Clinical Terms is a common clinical language consisting of clinical phrases or terms that can be grouped together with relationships between them; hence it provides a flexible coding system for rare disease.
Specialised/highly specialised	<p>Those services that require a level of clinical expertise that may not be available in every hospital. Highly specialised services require a level of clinical expertise that means they can only be delivered in a very small number of expert centres, usually no more than three in England.</p> <p>In this document, the term ‘specialised and ‘highly specialised’ refer to the commissioning models used by NHS England to commission specialised and highly specialised services respectively.</p>
Specialist/highly specialist	In this document, the term ‘specialist’ refers to a level of expertise delivered within a clinical service (with ‘highly’ specialist meaning a very high level of expertise).
UK Genomic Knowledge Base	The growing sum of knowledge being accrued as a result of the 100,000 Genomes Project when taken together with the existing research base.

<sup>1</sup> <https://www.gov.uk/government/publications/rare-diseases-strategy>

<sup>2</sup> <https://www.england.nhs.uk/five-year-forward-view/>

<sup>3</sup> <http://www.legislation.gov.uk/2017/296>

<sup>4</sup> <https://www.england.nhs.uk/commissioning/spec-services/highly-spec-services/>

<sup>5</sup> <https://www.england.nhs.uk/commissioning/rdag/>

<sup>6</sup> <https://www.gov.uk/government/groups/uk-rare-disease-forum>

<sup>7</sup> <https://www.raredisease.org.uk/media/1588/the-rare-reality-an-insight-into-the-patient-and-family-experience-of-rare-disease.pdf>

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- <sup>8</sup> <https://healthmedia.blog.gov.uk/2017/07/04/cmo-annual-report-on-generation-genome/>
- <sup>9</sup> <https://www.genomicsengland.co.uk/>
- <sup>10</sup> <https://www.gov.uk/government/publications/developing-a-consensus-on-data-sharing-to-support-nhs-clinical-genetics-and-genomics-services>
- <sup>11</sup> <https://www.raredisease.org.uk/our-work/rare-disease-care-coordination-delivering-value-improving-services-2013/>
- <sup>12</sup> <https://www.geneticalliance.org.uk/news-events/news/hidden-costs-report/>
- <sup>13</sup> <https://www.judiciary.gov.uk/publications/paul-ashton/>
- <sup>14</sup> <https://www.raredisease.org.uk/our-work/patient-experiences-of-transition-between-care-providers-2014/>
- <sup>15</sup> <https://www.england.nhs.uk/publication/nhs-england-board-meeting-papers-30-november-2017/>
- <sup>16</sup> <https://www.england.nhs.uk/wp-content/uploads/2017/01/specialised-participation-frmwrk.pdf>
- <sup>17</sup> <https://www.england.nhs.uk/publication/methods-national-service-specifications/>
- <sup>18</sup> <https://www.england.nhs.uk/publication/methods-national-clinical-policies/>
- <sup>19</sup> <https://www.england.nhs.uk/publication/specialised-commissioning-service-development-policy-and-process/>
- <sup>20</sup> <https://www.england.nhs.uk/nhs-standard-contract/17-19-updated/>