

## UK STRATEGY FOR RARE DISEASES - COMMITMENTS

(lead organisation as of February 2014)

Number	Commitment	Organisations leading on action in England (seeking contributions from other organisations as appropriate)
1	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.	All relevant organisations in line with their remit
2	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.	All relevant organisations in line with their remit
3	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.	NHS England
4	Make sure that patients and their families have a say in decisions about treatment and in the planning, evaluation and monitoring of services.	NHS England
5	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.	NHS England
6	Improve access for patients (or where appropriate their parents or guardians) to their personal data.	NHS England
7	Support patients to register on databases, where these exist.	Public Health England/NHS England

Number	Commitment	Organisations leading on action in England (seeking contributions from other organisations as appropriate)
8	Help patients to contribute to research and other activity related to rare diseases.	NHS England/National Institute for Health Research
9	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.	DH/UK National Screening Committee
10	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.	NHS England
11	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 e.g. to help disabled children who are thought to have a genetic syndrome or condition that science has not yet identified.	NHS England

Number	Commitment	Organisations leading on action in England (seeking contributions from other organisations as appropriate)
12	<p>Work with the NHS and clinicians to establish appropriate diagnostic pathways which are accessible to, and understood by, professionals and patients, by</p> <ul style="list-style-type: none"> <li>— establishing clear, easily accessible and effective pathways between primary care, secondary care, regional centres and specialist clinical centres, as appropriate</li> <li>— putting protocols in place to identify patients with no diagnosis, ensuring that a lack of diagnosis does not create a barrier to treatment</li> <li>— drawing on patients' ability to help inform decisions about referral and diagnosis</li> <li>— creating effective clinical networks to support this process</li> <li>— making high quality diagnostic tests accessible through common, clinically agreed systems or pathway</li> <li>— embedding appropriate information in national data systems including measuring equity of access to molecular tests to maintain UKGTN diagnostic studies</li> </ul>	NHS England
13	Ensure that there are appropriate procedures for evaluating the costs and benefits of treatments for patients.	NICE
14	Where appropriate, support the availability of computerised prompts to help GPs diagnose a rare disease when a rare disease has not previously been considered.	NHS England
15	<p>Improve education and awareness of rare diseases across the healthcare professions, including:</p> <ul style="list-style-type: none"> <li>— involving patients in the development of training programmes</li> <li>— encouraging medical, nursing and associated health professionals to get hands-on experience in specialist clinics</li> <li>— ensuring awareness of methods and clinical techniques used in differential diagnosis.</li> </ul>	Health Education England

Number	Commitment	Organisations leading on action in England (seeking contributions from other organisations as appropriate)
16	Monitor the development of ICD-11 in preparation for its adoption.	All relevant organisations in line with their remit
17	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
18	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 to establish responsibility
19	Explore options to improve the link between existing patient data and electronic health records.	NHS England is currently in discussion with relevant agencies to establish responsibility
20	Assess the potential for rare disease databases where they do not exist.	Department of Health
21	Agree international standards, building on existing NHS standards.	NHS England
22	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

Number	Commitment	Organisations leading on action in England (seeking contributions from other organisations as appropriate)
23	<p>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:</p> <ul style="list-style-type: none"> <li>— an appropriate care plan for all patients with a rare disease</li> <li>— clearly stated principles around the standards of care which patients with a rare disease can expect, including patients with no diagnosis</li> <li>— the development of seamless pathways for transition, from childhood to adolescence, and on to adulthood and older age</li> <li>— access criteria and measures of quality and outcomes</li> </ul>	NHS England
24	<p>Agree that specialist clinical centres should as a minimum standard:</p> <ul style="list-style-type: none"> <li>— have a sufficient caseload to build recognised expertise</li> <li>— where possible, not depend on a single clinician</li> <li>— coordinate care</li> <li>— arrange for coordinated transition from children's to adults' services</li> <li>— involve people with rare conditions, and their families and carers</li> <li>— support research activity</li> <li>— ensure their expertise is available to families and their healthcare teams</li> </ul>	NHS England
25	<p>Ensure that the relationship between the specialist clinical centres and science and research is explained to and understood and put into practice by: practitioners delivering local health and social care; the research community; industry; academia.</p>	NHS England/Department of Health
26	<p>Set out clearly the connections to and communications with specialist clinical centres in molecular diagnostics and other forms of diagnostic support.</p>	NHS England/Department of Health

Number	Commitment	Organisations leading on action in England (seeking contributions from other organisations as appropriate)
27	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 ultra-rare).	NHS England/Department of Health
28	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.	NHS England
29	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.	Public Health England/NHS England
30	Identify how they can change systems to hold information about rare diseases, including information about the uptake of treatments.	NHS England
31	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.	Department of Health
32	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
33	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.	NHS England

Number	Commitment	Organisations leading on action in England (seeking contributions from other organisations as appropriate)
34	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
35	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.	Department of Health/NHS England
36	Encourage patient groups to get involved with regulatory bodies.	Department of Health
37	Help patient organisations and community engagement events develop more formal partnerships with the NHS research-active organisations.	NHS England/National Institute for Health Research
38	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.	National Institute for Health Research
39	Work with the research community, regulators, providers of NHS services and research funders to develop risk-proportional permission systems.	Health Research Authority
40	Encourage researchers to use current guidance to produce generic participant information leaflets and consent forms and participate in future guidance reviews.	Health Research Authority
41	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.	National Institute for Health Research

Number	Commitment	Organisations leading on action in England (seeking contributions from other organisations as appropriate)
42	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.	UK Genetic Testing Network/NICE
43	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	UK Genetic Testing Network/NHS England/NICE
44	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/Health Education England
45	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.	Health Education England/NHS England/Genomics England Limited
46	Work with industry to set priorities and determine how best to support research into rare diseases and promote research collaboration.	National Institute for Health Research
47	Support initiatives to facilitate engagement between patients, clinical care teams, researchers and industry wherever practical.	NHS England/National Institute for Health Research
48	Set out the benefits of collaboration (besides producing specific treatments) for all stakeholders.	NHS England/National Institute for Health Research



Number	Commitment	Organisations leading on action in England (seeking contributions from other organisations as appropriate)
49	Continue to build a cohesive infrastructure for implementation and coordination of rare disease research in the NHS.	National Institute for Health Research
50	Encourage major research funders to use current structures to coordinate strategic funding initiatives in rare diseases.	Department of Health [through its membership of the Office for Strategic Coordination of Health Research (OSCHR) Board and the UK Clinical Research Collaboration (UKCRC)]
51	Improve engagement between key stakeholders, including: <ul style="list-style-type: none"> <li>— patients and relatives</li> <li>— main funding providers</li> <li>— healthcare commissioners</li> <li>— NHS hospitals and specialist care units</li> <li>— industry (pharmaceutical, biotechnology, IT, diagnostics)</li> </ul>	NHS England/National Institute for Health Research