

Consultation on the United Kingdom Plan for Rare Diseases

February 2012



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Contact details	Sarah Bramley-Harker Screening & Specialised Services Team Department of Health, England Quarry House, Quarry Hill, Leeds LS2 7UE 0113 254 6708 rarediseasesconsultationresponses@dh.gsi.gov.uk
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1 Foreword

1. On 11th November 2008, the European Commission published a Communication and Proposal for a European Council Recommendation on Rare Diseases. The Recommendation, which aims to introduce measures at European level to combat rare diseases, was accepted in 2009. This consultation document and responses will form the basis of the UK's Plan on Rare Diseases in response to the Recommendation.
2. Rare diseases can affect anybody at any stage of their life and people being treated for rare diseases need support from a range of professions including general practitioners, diagnostics staff, the specialist nurses and clinicians providing their care, social care and, of course, their friends and family. This consultation document recognises this and is deliberately broad in order to cover the range of expertise that may be needed.
3. This document has been developed jointly by the four nations of the UK and sets out a coherent and joined up approach to tackling rare disease. We recognise the challenges faced by people with a rare disease and we hope that this document will help to address some of the issues highlighted.
4. For example, we know that more needs to be done to raise the profile of rare diseases. As advances in science and technology are enabling more children with rare diseases to live into adulthood we need to improve the transition from children's services to adult services. The current systems for coding hospital activity and mortality do not serve rare disease well but we are optimistic that the next revision of the International Classification of Disease due in 2015 will help.
5. Many rare diseases are of genetic origin and it is vital that we embrace the advances in genetics and genomic medicine and ensure that the NHS is ready to take advantage of these developments. We need to reduce diagnostic delay and ensure that training for all clinicians enables them to be alert to the possibility of a rare disease in their patients and to be able to direct patients accordingly for diagnostic investigations.

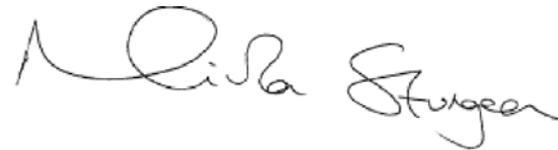
6. As Ministers of the four UK Health Departments we are committed to improving services for people with rare diseases and hope that through this consultation document and the Plan we can significantly raise their profile, leading to better outcomes for the many patients and their families/carers who are affected.



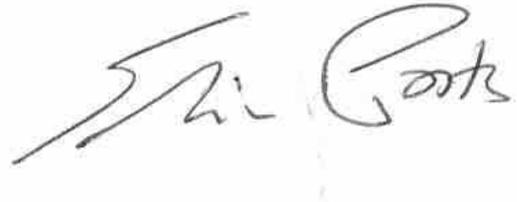
Earl Howe
Parliamentary Under Secretary of
State for Quality
England



Lesley Griffiths AM
Minister for Health and
Social Services
Wales



Nicola Sturgeon MSP
Cabinet Secretary for Health,
Wellbeing and Cities Strategy
Scotland



Edwin Poots MLA
Minister for Health, Social Services
and Public Safety
Northern Ireland

2 Executive summary

7. Rare diseases can affect physical or mental health or both. More than 5,000 rare diseases have been identified.
8. Diagnosis of a rare disease is difficult and delays may occur. Sometimes it is obvious that there is a problem, but expertise is needed to establish the precise diagnosis. Clear referral pathways to expert centres can help in this situation. But sometimes a rare disease is not even suspected. In this situation, computer prompts for GPs, similar to those already used to alert GPs to issues in prescribing, are a promising avenue to explore.
9. Screening at birth for some rare diseases is another way to avoid delay in diagnosis. There are already national screening programmes for several rare diseases. The United Kingdom National Screening Committee advises Ministers and the NHS in the four United Kingdom countries on all aspects of screening.
10. Training of medical and other staff in the NHS is among the best in the world, but we must not be complacent. We need to ensure, through both basic training and continuing professional development, that all doctors are alert to the possibility of a rare disease when they see a patient.
11. Coding and classification issues are important if we are to understand fully the burden of rare disease. The current, 10th, version of the International Classification of Disease (ICD 10) does not serve well for rare disease. A new revision is expected in 2015 which should resolve this problem.
12. The United Kingdom is at the cutting edge of international research in rare disease. Most of the biomedical research centres funded by the National Institute for Health Research (NIHR) are conducting research on rare disease. The UK participates in rare disease research at European level and will be actively involved in the new International Rare Disease Research Consortium (IRDiRC).

13. There is a consensus among experts and people with rare conditions that expert centres caring for people with rare and complex disorders must have the following characteristics:
 - co-ordinated care;
 - adequate caseload for expertise;
 - not dependent on a single clinician;
 - arrangements for transition from children's to adults' services;
 - engaged with people with rare conditions;
 - research active.
14. Networks are a crucial part of any system of care for rare disease. Two types of network are required. Firstly, expert centres must network with each other to devise protocols, agree consensus documents, collaborate in research and seek each other's advice on particular patients. The second type is the network of local hospitals which refer patients to the expert centre. Each expert centre must know which local hospitals it has responsibility for, and the local hospitals must know which expert centre they can look to for help, support, advice and assistance. This does not preclude patient choice but strong links between each centre and its referring hospitals are the foundation of good co-ordinated care.
15. In order to stimulate the development and marketing of new drugs for rare diseases, the European Union offers a range of incentives. The UK takes an active role in the decision making process for this 'orphan medicine' arrangement. Where effective new medicines become available for the treatment of rare and very rare conditions, patients are, understandably, concerned that they should have access to the drugs that their doctors recommend. Our priority is to give NHS patients better access to effective and innovative medicines and that is why we are working towards a new system of pricing for branded medicines, where the price of a drug will be linked to its assessed value.
16. Expert clinical teams can offer advice and treatment but the real experts in living with a rare disease are of course the people suffering from the disease and their families. In the United Kingdom there is a wide range of patient organisations which offer help and support to people with rare disease.

17. Information is key to patient empowerment. Newly diagnosed patients and their families need information in a variety of languages and formats, and the information must be supplied at a pace that people struggling to come to terms with often devastating news can understand. Personalised care plans are an important element in the care of any patient with a long-term condition. This empowers patients and their carers and ensures a coherent approach from all those involved in the care plan.
18. Information is widely available on the internet but quality is variable. The UK participates in the Orphanet website, which is quality assured and one of the most comprehensive websites on rare disease in the world. The NHS also provides information on a variety of websites such as NHS Inform, NHS Evidence and NHS Choices.
19. Registers are a key tool in the struggle to understand rare disease. They are key to high quality care despite the fact that they have often regarded as the domain of researchers but people with rare disease see value in being included on a register. It is the route to trials of new therapies, but also gives people a hope for the future and the comfort that they are not forgotten and not alone.
20. A plan must be sustainable and affordable within existing resources.

3 Introduction

21. The United Kingdom can be proud of its record in treating people with rare disease. We have world-class research teams at the forefront of discovery and innovation in the treatment of rare disease. We have dedicated teams of clinicians and healthcare staff who provide care for complex conditions equal to the best available anywhere in the world, and we have strong patient organisations that powerfully articulate the needs and priorities of people with rare disease. However, there is always room for improvement.
22. There are already a number of initiatives in place across the UK to support people with long-term conditions, including those where the condition is a rare disease. For example in England the Long Term Conditions National Service Framework aims to give people more choice and support through services planned and delivered around their individual needs. In Scotland, the Scottish Government's strategic partnerships with the Long Term Conditions Alliance Scotland (LTCAS) and the Neurological Alliance Scotland, have an important role in helping the voluntary sector shape the development of services for people living with long-term conditions, including rare diseases. In Northern Ireland, support to people with long term conditions will be provided under the Physical and Sensory Disability Strategy for 2012 – 2015, the Long Term Conditions Policy Framework (due for publication later in 2012) and NI Executive response to the Bamford Review (which encompasses mental health and learning disability) and associated implementation plans. In Wales, implementation of the Chronic Conditions Management programme of work (2008-11) introduced a proactive and managed approach to managing long term conditions based upon early assessment and diagnosis and appropriate treatment within the community.
23. In 2009, the European Council published a Recommendation on an action in the field of rare diseases. This Council Recommendation asked every member state of the European Union to develop a national plan or strategy, preferably by the end of 2013, on rare diseases. The United Kingdom supported this Recommendation.

24. Health Services are organised and managed separately in each of the four nations of the United Kingdom. All four have agreed to work together on a single response to the Council Recommendation and this consultation represents the result of that collaboration, offering a framework for managing rare diseases wherever they occur. Each nation will then take forward the Plan in line with its own priorities and patterns of service engaging with other nations as necessary.

3.1 The economic context

25. The NHS will continue to develop new treatments and ways of working for people with rare conditions. The current economic and fiscal environment is very challenging. Improvements and developments to services are needed which are sustainable and realistic within existing resource levels which also reflect the priorities of the four nations of the United Kingdom.
26. Many improvements in the care of people with rare disease will save the NHS money:
 - a recent unpublished study suggests that better care of patients with rare neuromuscular diseases could save the NHS £31m per annum by avoiding emergency admissions;
 - effective treatment of neuromyelitis optica, a rare neurological condition, reduces the number of disabling relapses, which are expensive to treat, by 80%. In England, establishing expert centres has substantially reduced the delay in diagnosis and start of effective treatment.
27. These examples illustrate the point that many improvements can be made that will not require extra funding. Moreover, a more coordinated approach may speed diagnosis, improve patient experience and outcomes, and save resources resulting from duplication of effort and waste. This can be illustrated by a recent case where a baby was seen by a health visitor with pink-stained nappies, a GP with 'eczema', a paediatrician with 'failure to thrive', an endocrinologist with excessive hair growth and a dentist with tooth discolouration. It was an expert dermatologist who, by diagnosing the rare disorder congenital erythropoietic porphyria as the cause of all the apparently unrelated problems, enabled all this effort to be better focused with a far better use of the time and energy of those treating the case. Better collaborative working should also lead to improvements in awareness of and advocacy for rare diseases; not only in the general population, but also in healthcare professionals and in policy makers.

28. This consultation will run from 29th February until 25th May 2012. The consultation document broadly follows the format of the European Council Recommendation on Rare Diseases and provides information about the systems currently in place within the UK for treating people with rare diseases. It also recognises that there are things that we could do better and invites comments on our suggestions for improvements that could be made in a number of areas. We would particularly welcome comments on the specific questions set out in this consultation. A consultation reply form is provided at the end of this document.

3.2 The problem of rare disease

29. The EU definition of a rare disease is one that affects fewer than 5 in 10,000 of the general population. Rare diseases can affect physical or mental health or both. They can range from debilitating, life-limiting conditions to manageable conditions that do not affect daily living. Around 80% of rare diseases are genetic.
30. The problems that arise in relation to rare diseases are well understood in the United Kingdom. For example, in 2009 the Chief Medical Officer for England highlighted the problem of rare diseases in his Annual Report:
- although individually these diseases are rare, collectively they are a large part of the work of the National Health Service;
 - rare diseases are very difficult to diagnose, treat and manage. This can lead to misdiagnoses and unnecessary treatments, which wastes NHS resources;
 - awareness and understanding of rare diseases across the health system can be poor, leading to difficulties in co-ordination and communication. This results in delays in diagnosis and treatment;
 - the majority of people affected by rare diseases are children. More and more children with rare diseases are surviving into adulthood because of improved treatments and services, but adult services for rare diseases are often lacking.
31. The four nations of the United Kingdom have separate health systems, but their guiding principles are the same. The four Health Ministers have reaffirmed their commitment to the core principles on which the NHS was founded. They affirmed that:
- the NHS provides a comprehensive service, available to all;

- access to its services is based on clinical need not an individual's ability to pay;
 - the NHS aspires to high standards of excellence and professionalism;
 - NHS services must reflect the needs and preferences of patients, their families and their carers;
 - the NHS works across organisational boundaries with other organisations in the interests of patients, communities and the wider population;
 - the NHS is committed to providing the best value for taxpayers' money, making the most effective and fair use of finite resources;
 - the NHS is accountable to the public, communities and patients that it serves;
 - everyone counts. Resources will be used for the benefit of the whole community to make sure that nobody is excluded or left behind.
32. These principles apply to all, no matter whether the condition a person suffers from is common or rare.
33. This consultation on the plan for rare diseases covers the four nations of the United Kingdom and aims to improve co-ordination and co-operation, achieve better outcomes, strengthen research and monitoring activities, engage and empower patients and their families or carers, and raise awareness in the public and in professionals.
34. Northern Ireland is unique in that it shares a land border with another member state of the EU. The accessibility of services in terms of distance combined with the high standard of transport links, means it may be preferable to develop clinical networks with the Republic of Ireland as well as Great Britain to support sustainable service models in Northern Ireland. Hence there is the need for some flexibility of arrangements and collaboration where possible in the interests of patients.

35. Additionally, the role of genetics in both the origins of rare disease and in their treatment is a key factor. This was emphasised by the Prime Minister's Council for Science and Technology in their report published in September 2011¹ which said:

“Recent developments in genome science and human genetics are on the threshold of delivering new diagnostic methods and innovative products to the clinic...Genetics will play a crucial role in the future in identifying drug targets and stratifying patient populations for the commercial sector.” This theme, and its implications for innovation in the NHS and the UK life sciences sector, are being taken forward as a key plank of the Autumn Package on Life Sciences announced by the Prime Minister on 5 December”.

3.3 Public awareness of rare disease

36. The European Commission published a report in 2011 on the “European Awareness of Rare Diseases”. The data in this study were collected by face-to-face interviews in each EU Member State. More than 1,200 interviews were conducted in the United Kingdom.
37. The results showed that public awareness of rare diseases is high in the United Kingdom. Most people understand what is meant by a rare disease, though only one in five of the United Kingdom respondents knew someone suffering from a rare disease. Almost all United Kingdom respondents (93 percent) agreed that people are unaware of the real problems faced by those suffering from a rare disease.
38. This survey also showed strong public support for managing rare diseases. Ninety eight percent totally agreed or tended to agree that people suffering from a rare disease need specific support from the NHS. This confirms the importance of developing this national plan.
39. In Europe, over 90 percent of all respondents were supportive of more European collaboration on rare diseases (including cross-border treatment) and the development of EU Member State plans or strategies on rare diseases.

¹ “The NHS as a Drive for Growth”, available at <http://www.bis.gov.uk/assets/bispartners/cst/docs/files/cst-reports/11-1096-nhs-driver-for-growth.pdf>

3.4 What people with a rare disease think

40. More coordinated action to address rare diseases is supported by bodies representing people with a rare disease.
41. Rare Disease United Kingdom (RDUK) is an alliance of key stakeholders brought together to develop strategic planning for rare diseases in the United Kingdom. It is a joint initiative of Genetic Alliance United Kingdom (the national charity of over 130 patient organisations supporting all those affected by genetic disorders) and others.
42. During the summer of 2010 RDUK conducted a survey of patients' experiences. The survey received responses covering over 100 different diseases. The survey gathered information on patients' experiences of research, diagnosis, access to care, information, support and treatment.
43. Although there is much that is excellent in NHS services for people with a rare disease, key problems reported by participants in the survey include:
 - patients regularly suffer from delayed diagnoses and misdiagnoses;
 - a lack of information provided by the NHS on rare conditions;
 - patients lacked co-ordination of their care;
 - inequalities in treatments offered to patients around the United Kingdom.
44. The main conclusion of the report was that the United Kingdom needs a national strategy on rare diseases to reduce the fragmentation of services and to ensure that patients with rare conditions are able to access equitable high quality care, information and support in a timely manner.
45. RDUK published a further report in 2011 *Improving Lives, Optimising Resources: A Vision for the UK Rare Disease Plan* which sets out their recommendations for the UK's Plan on rare diseases. In their report RDUK call for more coherence in the way services for patients with rare diseases are designed and provided.
46. The Specialised Healthcare Alliance (SHCA) has outlined a strategy *Leaving no-one behind* aimed at improving the care of people with rare and complex conditions. The SHCA is a coalition of 71 patient related organisations, supported by ten corporate members, which campaigns on behalf of people with rare and complex conditions.

47. The report outlines a number of recommendations for specific services that are realistic and achievable without significant cost implications. These were derived from a series of multi-disciplinary workshops and inform a set of key generic drivers relevant to the delivery of cost-effective quality in specialised services.
48. The recommendations emphasise:
- the need for the doctor/ patient relationship to evolve in a way which speeds diagnosis through better referral pathways and the ability of many patients to help inform diagnosis, predicated on well signposted sources of reliable information;
 - the role of patient organisations in protecting the vulnerable and driving up standards, sometimes through the provision of more innovative services;
 - the need to ensure integrated pathways, especially in the development of multi-disciplinary networks;
 - the contribution of National Institute for Health and Clinical Excellence (NICE) quality standards or their equivalent to high quality specialised care and outcomes;
 - the desirability of outcome measures which capture both clinical and patient perspectives with a view to maximising effectiveness and efficiency;
 - the central importance of patient registries, with more sharply focused datasets to keep costs down.
49. The SHCA's report builds on earlier work on high quality specialised care by Sir David Carter in 2006.²

² Report of Sir David Carter on the "Review of arrangements for specialised services", May 2006

4 Speedy diagnosis and early intervention

4.1 Speedy diagnosis

50. Some rare disorders are instantly recognisable because of characteristic symptoms or signs, but others are more subtle. It can then take a long time to reach the correct diagnosis. Some diseases are so rare that an average GP will see a case once in a lifetime. So, although rare diseases are well covered in medical training in the United Kingdom, both at undergraduate and postgraduate level, it is unrealistic to expect GPs to recognise all rare diseases.

51. Five areas of difficulty for GPs in diagnosis were identified in a review carried out by Kostopoulou and colleagues in 2008:

- atypical presentations;
- non-specific presentations;
- very rare conditions;
- co-morbidity (more than one disease present);
- perceptual features that could be missed.

Often these features occur together, making the diagnosis of a rare disease particularly difficult. Three approaches may help to ensure speedy diagnosis in people with rare disease.

52. Firstly, clearly defined care pathways. Sometimes it is obvious that there is a problem, but an expert centre is needed to make an exact diagnosis. So, for example, if someone loses sensation in their hands and feet, a probable diagnosis is peripheral neuropathy. There are many common causes of peripheral neuropathy which can be established in local hospitals. However there are also some very rare causes which can only be fully identified with great expertise and comprehensive laboratories.

53. In this first situation clear pathways from primary care to local hospitals, and then to regional and national centres of expertise will help to speed diagnosis. The care pathways will be based on the care networks described below in section 7.

54. Secondly, a different strategy is needed to help speed diagnosis where a rare disorder is not even suspected. The problem here is that some rare disorders mimic common conditions. In this situation it may take months or even years before doctors become suspicious that a rare disease may be present.
55. In this second situation computerised databases and algorithms (the so-called 'expert systems') may help. A patient may have two symptoms which separately are each common but which only very rarely occur together. A computerised prompt could help to alert the doctor to this situation. Almost all GPs now routinely use computer systems in their clinics and surgeries. Computer prompts are already in common use to alert doctors to possible problems in prescribing, and a system developed by Surrey University is available to help GPs diagnose diabetes, including its rare forms, correctly (<http://www.clininf.eu/cod>).
56. Thirdly, other systems exist that can take advantage of extra information available when specialist doctors have examined the patient. For example, the SimulConsult system (<http://www.simulconsult.com/>) has been developed to help diagnosis in neurology and genetics, two fields of medicine with many rare diseases.

CONSULTATION QUESTION: Do you agree that commissioners of services should explore the potential of expert clinical systems to reduce diagnostic delay, particularly in neurology and genetics?

4.2 Training

57. Training of medical and other staff in the NHS is among the best in the world however we must not be complacent. The UK needs to ensure, through undergraduate and postgraduate training and continuing professional development, that all doctors are alerted to consider the possibility of a rare disease when they see patients. Curricula for the training of doctors are written by the Royal Colleges and approved by the General Medical Council.
58. Medical students and trainee doctors approach medical disease by eliciting symptoms and signs. The undergraduate and postgraduate curricula focus now on presentations of medical disease. The student or trainee doctor develops his/her diagnostic skills and how to access up to date information on medical illness and disease. Consequently, the undergraduate curriculum in UK medical schools includes education and training in the

diagnosis and treatment of rare disease as part of the differential diagnosis of presentations of medical conditions. Detailed knowledge is not expected, however a learning outcome is a general awareness of rare diseases presenting in all medical specialities.

59. Medical schools generally take an innovative approach to the teaching of skills relevant to rare disease. At Bristol medical school, for example, students are shown how to exploit the power of internet search engines to explore the possibility of a rare disease when a patient presents with an unexplained symptoms, or an unusual combination of symptoms. In the Peninsula Medical School students follow the patients' journeys in clinical weeks based on presentations of illness and will explore causes of relevant symptoms and signs with a differential diagnosis including rare diseases.
60. During speciality training at postgraduate level, a more detailed knowledge of the differential diagnosis of medical symptoms and signs is learnt. In general, while the curriculum for specialist training of doctors is developed to give more emphasis on common diseases, the curriculum focuses on signs and symptoms and the differential diagnosis consequently allowing an exploration of all causes of clinical presentations.
61. Time treating patients is often proportional to disease frequency since trainees learn from the patients they treat. Nonetheless, since trainees learn from a symptoms and signs approach to medical illness and learn how to access current information, the diagnosis of rare diseases is included in the syllabus.
62. After speciality training, all doctors embark on a programme of lifelong learning through continuing professional development. Centres of Excellence have a particular role to ensure, through a programme of lectures, publications and visits, that doctors are aware of new developments in the field of rare disease, including new treatments, new methods of diagnosis and the establishment of new expert centres. This activity helps to raise awareness of rare disease and so reduce diagnostic delay.

CONSULTATION QUESTION Can you suggest ways of rare disease featuring more prominently in speciality training for doctors?

4.3 Screening

63. The United Kingdom National Screening Committee, chaired by the Chief Medical Officer for Scotland, advises Ministers and the NHS in the four United Kingdom countries on all aspects of screening. Using research evidence, pilot programmes and economic evaluation, it assesses the evidence for programmes against a set of internationally recognised criteria covering the condition, the test, the treatment options and the effectiveness and acceptability of the screening programme. Assessing programmes in this way is intended to ensure that they do more good than harm at a reasonable cost. The United Kingdom National Screening Committee also sets up practical mechanisms to oversee the introduction of new programmes in the English NHS and monitors effectiveness and quality of these programmes.
64. The United Kingdom National Screening Committee regularly reviews policy on screening for different conditions in the light of new research evidence becoming available. All countries of the United Kingdom offer newborn screening for a set of rare diseases.
65. The newborn blood spot programmes test babies for a number of rare diseases. In England, this includes phenylketonuria, congenital hypothyroidism, sickle cell disease, cystic fibrosis and medium chain acyl CoA dehydrogenase deficiency (MCADD). Also the NHS Sickle Cell and Thalassaemia Screening Programme offers a linked programme of screening for sickle cell disease for all newborn babies in England and screening during pregnancy for all pregnant women in England.
66. In Scotland, all babies are offered screening for phenylketonuria, congenital hypothyroidism, cystic fibrosis, sickle cell disease and MCADD. Thalassaemia is screened for antenatally.
67. In Wales newborn screening is offered for hypothyroidism, cystic fibrosis and phenylketonuria. Screening is offered as part of routine care. Sickle cell and thalassaemia antenatal screening is offered for all pregnant women resident in Wales at an increased risk of having a child affected by a sickle cell disorder or thalassaemia major.
68. In Northern Ireland, universal screening of all infants at 5 days of age is offered for phenylketonuria, congenital hypothyroidism, cystic fibrosis and MCADD. Screening for homocystinuria and tyrosinaemia is also offered. Screening for sickle cell disease will commence in early 2012.

69. The policy process of the UK National Screening Committee includes regular review of existing programmes, and consideration of new programmes. The criteria used to assess proposals are set out on the Committee's website.
70. A core principle of United Kingdom screening programmes is that screening must have the potential to benefit the person who is subjected to the screening test. Proponents of wider screening point out that those benefits might accrue in the following ways:
- earlier diagnosis (e.g. at birth) may reduce the 'diagnostic odyssey' – the long trek suffered by some patients around many different doctors, having many different tests, before a correct diagnosis is finally reached. It can in some cases take months or years to make an accurate diagnosis of a rare condition. Earlier diagnosis may lead to more time being healthy;
 - screening for genetic disease allows parents to make informed choices about subsequent family planning, which may benefit the child with the disorder.
71. These potential benefits raise a number of ethical and other issues which require careful thought.

CONSULTATION QUESTION: Do you agree that the UK National Screening Committee should take into account the benefit of screening in reducing the 'diagnostic odyssey' and in allowing informed choice for subsequent family planning?

5 Coding and classification

72. The standard system for coding and classification of disease is the World Health Organisation's International Classification of Diseases, now in its 10th revision. This is known as ICD 10. It is used for national coding of death statistics, and for the coding of hospital activity throughout the NHS. Coding in the NHS is very accurate – a recent review showed that coding of primary diagnosis was accurate in 96% of cases in hospital statistics.
73. The ICD is a global standard which has been running in its various revisions for 100 years. It has proved its worth in the classification of common disorders. The ICD does not however serve so well for rare disease. There are two reasons for this.
74. Firstly, as science advances, new diseases (or subtypes of well known diseases) are constantly being described, so the classification is continuously becoming outdated. Secondly, the classification is explicitly designed to group the thousands of known disorders into a manageable number of categories. However, this grouping process can put together some rare and very rare disorders which we need to track separately.
75. So for example some babies are born with no ability to fight infection: severe combined immune deficiency (SCID). This used to be regarded as a single disorder but is now known to have many different causes, which require different treatment regimes. Some of the subtypes are treated in the NHS with novel therapies such as gene therapy, but the subtypes cannot all be identified in our routine data systems because they do not have separate codes.
76. As another example, ICD10 groups together in a single code a number of very rare diseases called sphingolipidosis. These are a group of severe inherited disorders: the current classification identifies seven subtypes.
77. The treatment of these separate diseases is very different – for two, enzyme replacement therapies are available; one is treated by stem cell transplant; and for some no specific treatment is yet available. One of the subtypes has its own subtypes A, B, C and D: effective drug treatment is available for type C but not for the other types. Again, none of these

complexities can be tracked in routine mortality or NHS data systems because of the way the coding system works.

78. The ICD has been modified in various ways to take account of complexities such as these, yet these modifications fail to separate out the rare and very rare disorders. A more comprehensive scheme has been produced by Orphanet, which is a rare disease project funded by the EU.
79. Meanwhile the ICD is being revised at the international level with the aim of publishing an 11th revision (ICD11) in 2015. The problems of coding for rare diseases are being addressed by a specific working party. Europe is at the forefront of this work, with the working party led by the Chair of the EU Committee of Experts on Rare Disease.
80. Changing the basic system of classification and coding requires a huge training exercise for medical coding staff in all NHS hospitals and in the national statistics organisations. Updated software is also needed for all computerised systems. So changing a fundamental coding system is not a task to be undertaken lightly.
81. Due to the likely cost it seems better to wait for the 11th revision of the ICD rather than attempt to introduce a new coding system throughout the NHS in the interim.
82. Other issues arise in computer systems which are not coded using the ICD. Examples include general practice (GP) systems and a number of laboratory systems. Advances in the branch of information science known as bio-ontology will help to ensure that these different coding systems work well together.

6 Research

83. The United Kingdom is at the cutting edge of international research in rare diseases. This includes research on the basic biological processes that cause rare diseases, the development of more effective ways of diagnosis and treatment, and research on patient experience. Research is carried out by universities, the NHS, and other organisations. It is funded by Government, in particular through the Medical Research Council and the National Institute for Health Research (NIHR), by medical research charities, the pharmaceutical, bioscience and healthcare industries, and other organisations including the European Commission.
84. In August 2011, the Government announced investment of £800 million over five years to develop NHS and university partnerships throughout England through the NIHR. The NIHR centres and units will help pull new scientific discoveries into benefits for NHS patients across a wide range of both major and rare diseases. Most of the current NIHR biomedical research centres of excellence are conducting research on rare diseases, and breakthroughs have included the world's first successful gene therapy for a rare and inherited form of blindness that causes progressive deterioration in vision and blindness in teenagers (Leber's congenital amaurosis). NIHR centres and units benefit from close links to basic research in the host universities funded by the Research Councils and charities, including research on disease mechanisms and relevant social science and ethics research.

CONSULTATION QUESTION: How can the NHS best ensure research in rare diseases carried out by the NIHR biomedical research centres and units is rapidly transferred into practice for the benefit of UK patients and their families and carers?

85. Research on rare diseases is also funded through other NIHR programmes and funding streams including the Health Technology Assessment programme, Research for Patient Benefit programme, the Clinical Research Network, and research training awards. The NIHR will continue to welcome funding applications for research into any aspect of human health including rare diseases. The Advisory Group for National Specialised Services works closely with the NIHR, and refers research

evidence for consideration within NIHR prioritisation processes. The Health Departments in Northern Ireland, Wales and the Scottish Government's Chief Scientist's Office also provide funding for research.

86. New technology for genome sequencing and rapid developments in understanding the human genome are likely to have a significant impact on research into the causes and diagnosis of rare diseases. The Research Councils and medical research charities have invested in centres providing UK researchers with access to the latest fast genome sequencing technology and in research programmes, such as the Wellcome Trust UK10K project to sequence 10,000 genomes, aimed at understanding the genetic influences on both common and rare diseases.
87. The relatively small number of patients affected by each rare disease presents methodological difficulties for evaluating new treatments and diagnostic techniques. This places additional importance on the value of international collaboration. The UK participates in rare disease research at a European level and has provided the lead for several collaborations within the Seventh Framework Programme (2007 – 13) including major projects on primary antibody deficiencies and on nuclease immune mediated brain and lupus-like conditions. The European Commission intends that rare diseases will continue to be a priority in the next research programme, Horizon 2020, which will run from 2014 – 2020.
88. The International Rare Disease Research Consortium (IRDiRC) was launched in April 2011 to foster international collaboration in rare diseases research. The NIHR has joined IRDiRC and will be actively involved as the Consortium pursues its goals to deliver new therapies and diagnostic tools for rare diseases.
89. In the *Plan for Growth* published with the Budget in March 2011, the Government made a commitment to publish plans for a secure data service focused on linking the data sets that do most to strengthen the international competitiveness of our life sciences research across major and rare disease areas. These plans were published in October 2011. The Clinical Practice Research Datalink will be established by 1st April 2012 and its functionality will be increased in stages over the next four years.

7 Centres of expertise and networks

7.1 Specialist centres

90. The management of rare diseases requires a particular concentration of health service expertise. Many rare diseases, particularly those of genetic origin, affect fundamental biological processes in every cell in the body – for example heart, liver, kidney – may be affected, demanding a range of specialist expertise. This expertise is required not just of the lead clinician but also of nurses, therapists, psychologists and diagnostic departments.
91. There is consensus among experts, commissioners and people with rare conditions that centres caring for people with rare and complex disorders, if they wish to be recognised as expert centres, must have the following characteristics:
- co-ordinated care;
 - adequate caseload for expertise;
 - not dependent on a single clinician;
 - arrangements for transition from children’s to adults’ services;
 - engaged with people with rare conditions;
 - research active.

Work is in progress towards ensuring that all expert centres in the UK have these characteristics.

92. *Co-ordinated care*: This matters where the disease, whether labelled “rare” or “genetic” or something else, affects a number of different organ systems. For example, there are many genetic causes of deafness, all rare. If the only problem is deafness, no special arrangements are needed. However, if the deafness is associated with renal failure, cardiomyopathy and diabetes then special arrangements are necessary.
93. Some cases may require expertise in a number of disciplines. For example the service caring for people with Biedl Bardet syndrome needs to call on diabetologists, ophthalmologists, nephrologists and all associated diagnostic and therapy services. Similarly, treating Alström syndrome will

need to call on cardiologists and transplant services. A different condition such as cystic fibrosis might require a different mix of specialist expertise. If a centre is treating all these simultaneously, it will need to be able to have all relevant specialities available.

94. With so many specialists, and so many hospital departments seeing the same patient to offer their particular expertise, co-ordinated care is essential. There have been examples of patients who have been required to attend the same hospital, often many miles from home, on several different days simply because the centre cannot co-ordinate their appointment properly into a single visit in a 'one-stop shop'.
95. The other requirement is full co-ordination between the expert centre and the local teams. Expert centres provide expertise but in almost all cases the bulk of care is provided locally – by local hospitals, by primary care teams, by social and education teams, and by the patient's family – so centres must have a good model for sharing their expertise with local providers. This requires written protocols for sharing care and constant communication between the expert centre and the local teams.
96. Local care is particularly important for people in remote areas, or where travel to the expert centre is difficult. For example patients from Northern Ireland may have to fly to centres in England. While this may often be the best solution other options may need to be considered including local care or, as noted earlier in this plan, links across the land border into the Republic of Ireland. In Scotland the Scottish Centre for Telehealth, which is now hosted in NHS 24, uses telemedicine to facilitate local care.
97. Patients can also help to coordinate their own care, particularly if empowered through a patient held record, which may be a booklet or an electronic record. Personalised care planning is recognised as a means of standardising excellent care for people with long term conditions and is another important element of coordinated care. Care planning:
 - puts the individual, their needs and choices that will support them to achieve optimal health and well-being at the centre of the process;
 - focuses on goal setting and outcomes that people want to achieve, including carers;
 - is planned, anticipatory and proactive with contingency (or emergency) planning to manage crisis episodes better (for those with complex needs);

- promotes choice and control by putting the person at the centre of the process and facilitating better management of risk;
- ensures that people, especially those with more complex needs or those approaching the end of life, receive co-ordinated care packages, reducing fragmentation between services;
- provides information that is relevant, timely and accredited to support people with decision making and choices (e.g. supported by an Information Prescription);
- provides support for self care so that people can self care/self manage their condition(s) and prevent deterioration;
- facilitates joined-up working between different professions and agencies, especially between health and social care; and
- results in an overarching, single care plan that is owned by the person but can be accessed by those providing direct care/services or other relevant people as agreed by the individual, e.g. their carer(s).

CONSULTATION QUESTION: Do you agree that commissioners of care for people with rare diseases should assess options for improved care co-ordination, including named care coordinators?

98. Accurate diagnosis of rare disease often demands great expertise in imaging, pathology and laboratory testing. Quality in NHS pathology and laboratory services is assured both by external accreditation of laboratory systems, for example by Clinical Pathology Accreditation (CPA) or other accreditation providers, and also by external quality assurance schemes for particular tests. For example in Scotland the Molecular Genetics and Cytogenetics Laboratory Consortium is well regulated and all four sites are CPA accredited. All tests provided have had gene dossiers approved by the United Kingdom Genetic Testing Network.
99. *Adequate caseload for expertise*: It is important for expert centres to see enough people with a particular rare disease to build and maintain expertise in all members of the team. This implies a limit to the number of centres. People with a rare disease frequently say they are happy to travel to receive expert care from teams knowledgeable in all aspects of their condition, particularly if this is balanced by the good shared care described above.

100. *Not dependent on a single clinician* A robust service cannot depend on a single clinician. Services for patients with rare disease must be available 24/7, at weekends, during holidays and so on. Resilience requires more than one individual to keep the service running at all times, responsive to patient need.
101. *Arrangements for transition from children's to adults' services*: There is an increasing need to provide for children with complex disorders who are surviving to adulthood. It is not helpful if this transition is associated with a complete disruption of care. All paediatric services need clear plans for transition from children to adult services. Linked groups of health professionals working together in a co-ordinated way through "managed clinical networks" can help and so can clear care pathways.
102. In Northern Ireland the DHSSPS launched the document *Developing Services to Children and Young People with Complex Healthcare Needs*, which highlighted service developments to be taken forward under the aegis of a Regional Inter-agency Implementation Group (RIIG). The role of this group was to drive and co-ordinate the regional implementation process at commissioning and Trust level. One of these service developments was the area of transitional care for these children into adult services. A reference group was set up to address these issues.
103. *Engaged with people with a rare disease*: Specialised centres for rare disease have a responsibility for empowering people with a rare disease and their families, including active engagement with patient organisations.
104. *Research active*: People with a rare disease, particularly those with untreatable disorders, and their families place a very high priority on research. Sometimes this is because they want to access the latest trials of promising treatments. However, even if their disease is as yet untreatable, people with rare disease place a high priority on accumulating more knowledge about the condition. So disease centres need to be fully engaged in research which will almost invariably involve co-operation with other centres across the United Kingdom and beyond.
105. Services which meet these requirements should be commissioned to provide specialised care for patients with rare diseases, as part of a clinical network at national, regional and local level, where possible.

CONSULTATION QUESTION: Do you agree that this list of criteria for expert centres should be the basis for future shaping of services:

- **co-ordinated care;**
- **adequate caseload for expertise;**
- **not dependent on a single clinician;**
- **arrangements for transition from children's to adults' services;**
- **engaged with people with rare conditions;**
- **research active?**

106. All of this means that special arrangements are needed to plan, fund and monitor (i.e. commission) services for people with rare disorders. These arrangements have existed in the United Kingdom for over 20 years with centres being “designated” for the treatment of particular rare conditions to facilitate a concentration of clinical expertise.
107. Subject to the passage of the Health and Social Care Bill through Parliament, in England, specialised services will in future be the responsibility of the NHS Commissioning Board, mandated by ministers. This will ensure that all patients can access equitable high quality services, regardless of which rare condition they have and regardless of where they live. It will ensure comprehensive arrangements for all rare diseases and is expected to end any suggestion of a ‘postcode lottery’. The NHS Commissioning Board will work with local commissioners of care, including social services, to ensure co-ordinated and joined up pathways for those with a rare condition. Working through local Health and Wellbeing Boards will be key to this joined-up commissioning.
108. In Scotland the National Services Division has commissioned specialised services along with genetic tests (either on a Scotland basis or through arrangements with England) and has been able to achieve huge efficiencies by doing so along with ensuring that services are available for people with rare disorders.
109. Similar arrangements exist in Wales under the Welsh Health Specialised Services Committee; and in Northern Ireland.
110. These arrangements provide a solid foundation for the care of people with rare disease throughout the United Kingdom.

7.2 Networks

111. Networks are a crucial part of any system of care for rare disease. The word 'network' can refer to two very different things and this is a source of confusion.
112. Firstly, networks of expert centres sharing good practice. If more than one expert centre exists to treat a rare condition we would expect those centres to meet regularly, to devise protocols, to agree consensus documents, to collaborate in research and to seek each other's advice on particular patients. These can often be international networks.
113. The European Commission has funded a number of pilot projects over the past 10 years in their programme of fostering networks of centres of expertise, also known as reference networks. The pilot projects have been very diverse in purpose and content. A particularly successful example, the Dyscerne project, was based in Manchester. Dyscerne created a network of expert centres across Europe to aid the diagnosis of dysmorphologies – the genetic conditions which affect the facial appearance of babies and children.
114. Secondly, hospitals refer patients to a specialised centre as part of a local network. Each expert centre must know which local hospitals it has responsibility for, and the local hospitals must know which expert centre they can look to for help, support, advice and assistance. This does not preclude patient choice, but strong links between each centre and its referring hospitals are the foundation of good co-ordinated care.
115. This second type of network is also the foundation of prompt care pathways and speedy diagnosis.

CONSULTATION QUESTION: Do you agree that each expert centre must know its network of local hospitals, and the local hospitals must know the pathway to the expert centre which will offer help, support, advice and assistance?

7.3 Genetic testing

116. Exact diagnosis of a rare disorder may require taking a family history and doing a genetic test. The United Kingdom Genetic Testing Network (UKGTN) advises the NHS on genetic testing across the whole of the United Kingdom. It aims to ensure the provision of high quality equitable

genetic testing services. The network is a collaborative group of genetic testing laboratories, clinicians and commissioners of NHS genetic services and involves patient support groups.

117. A gene dossier is completed by a testing laboratory and the specialist who wishes to provide a new genetic test. The gene dossier is evaluated as a part of a process to ensure new test methods meet the quality standards of the NHS.
118. The UKGTN will assess whether providing a test as an NHS service is likely to be of benefit to patients. If the test is approved, the recommendation will be passed to the national Genetics Commissioning Advisory Group (GenCAG) who will request that the relevant health commissioners consider providing funding for the test. Those tests that are approved by GenCAG are listed in the Directory of Molecular Genetic Testing.
119. This system ensures that doctors know which tests are available where, and at the same time helps to avoid wasteful duplication of effort. This system works well and will be retained.
120. The range and scope of genetic tests has increased dramatically in recent years, and it is hard for all doctors to stay up to date with latest developments. It is therefore essential that when a laboratory report is issued, the report also includes for the clinician who requested the test advice on who to contact for further information.
121. More directly a positive test result from a laboratory should include a recommendation on where the patient should be referred for further investigation and expert care. Clinicians who order tests cannot be expected to know the expert centre for every rare disease which the laboratory may diagnose. So, information from the laboratory is useful. This reinforces the importance of clear care pathways and designated centres for rare disease.

7.4 Medicines for rare diseases

Current system

122. Patients with rare diseases deserve the same quality, safety and efficacy in medicines as other patients with more complex conditions. However, for rare diseases the cost of developing and bringing a drug to market may not

be recovered by expected sales. In order to stimulate the development and marketing of drugs for rare diseases, the European Union offers a range of incentives. In particular, after the granting of a marketing authorisation, orphan medicinal products benefit from 10 years market exclusivity in the EU. During that period, directly competitive similar products cannot normally be placed on the market.

123. Applications for the designation of orphan medicines are reviewed according to Regulation (EC) No 141/2000 by the European Medicines Agency, through the Committee for Orphan Medicinal Products (COMP). Via the Medicines and Healthcare products Regulatory Agency (MHRA), the UK takes an active role in the decision making process at the COMP, ensuring applications for Orphan Drug designation of potential drug candidates for rare diseases are appropriately recognised, encouraging companies to develop their products further.
124. For orphan designation, the following criteria must be fulfilled:
- the medicinal product is intended for the diagnosis, prevention or treatment of a life-threatening or chronically debilitating condition affecting no more than 5 in 10,000 persons in the European Union or without incentives it is unlikely that expected sales of the medicinal product would cover the investment in its development;

and

 - no satisfactory method of diagnosis, prevention or treatment of the condition concerned is authorised, or, if such method exists, the medicinal product will be of significant benefit to those affected by the condition.
125. For drug licensing, it is compulsory for designated orphan medicinal products to use the centralised procedure to gain a marketing authorisation. A marketing authorisation application will be sent by the company directly to the European Medicines Agency, to be assessed by the Committee for Medicinal Products for Human Use (CHMP). The UK is fully represented at the CHMP, ensuring that applications for a Marketing Authorisation (MA) for an Orphan Drug are thoroughly and rapidly evaluated for quality, safety and efficacy and a MA is granted without undue delay for the treatment of rare diseases. The procedure results in a Commission decision, which is binding on all EU Member States.

126. Where effective new medicines become available for the treatment of rare and very rare conditions, patients are, understandably, concerned that they should have access to the drugs that their doctors recommend. Our priority is to give NHS patients better access to effective and innovative medicines and that is why we are working towards a new system of pricing for branded medicines, where the price of a drug will be linked to its assessed value.

Future arrangements

127. The Government will introduce value-based pricing from January 2014, when the current pricing arrangements (the Pharmaceutical Price Regulation Scheme 2009, “PPRS”) for branded medicines comes to an end. Like the PPRS, it is expected that value-based pricing will be a UK-wide system. Proposals for the new system were set out in the consultation document *A new value-based approach to the pricing of branded medicines*³ in December 2010. The consultation closed on 17 March 2011, and the Government response⁴ to the consultation was published on 18 July 2011. There is more work to do to develop the new arrangements including engagement with patients, including representatives of patients with rare diseases, clinicians, the NHS, taxpayers, industry and other interested parties as our work progresses.
128. Equal consideration should be given to patient safety through active surveillance of adverse drug reactions for orphan medicines performed at selected sentinel sites, through drug event monitoring, the use of registries and by comparative observation studies. Such activities in respect of orphan medicines should be aligned the remit of the new Pharmacovigilance Risk Assessment Committee (PRAC), expected to come into function in 2012 under EU pharmacovigilance legislation (Directive 2010/84/EU).
129. Unlicensed or off label medicines may be considered for patients with rarer conditions as well as for patients with common conditions which are hard to treat and where there is no licensed medicine that is clinically appropriate. At a local level, the NHS often does not have the evidence it needs to make an informed decision about these medicines. So, following a successful pilot by the UK Medicines Information Service which looked at providing this information, the Department of Health in England have recently asked the National Institute for Health and Clinical Excellence

3 http://www.dh.gov.uk/en/Consultations/Closedconsultations/DH_122760

4 http://www.dh.gov.uk/en/Consultations/Responsestoconsultations/DH_128226

(NICE) to take this on. NICE will ensure a high quality service is provided that summarises the best available evidence for selected unlicensed and off-label medicines where a demand for information has been identified in the NHS in England. This new service will provide information aimed at decision-makers and patients in the NHS to inform local prioritisation, treatment and funding decisions in circumstances where there are no clinically appropriate licensed alternatives. It is expected that the new service will begin in the spring of 2012.

8 Empowering those affected by rare conditions

130. Expert clinical teams can offer advice and treatment but the real experts in living with a rare disease are of course the people suffering from the disease and their families.
131. In the United Kingdom there are a wide range of patient organisations which offer help and support to people with rare disease. Most offer advice and support through websites, leaflets and by personal contact. In the early stages of diagnosis it is enormously helpful to talk to other people with the same condition and families for practical and emotional support. This ranges from tips and hints on how to use dressings or take a bath through to guidance on what benefits and services are available.
132. In Northern Ireland, the DHSSPS and all health and social care bodies have a statutory duty to promote public involvement and consultation including a duty to promulgate information about health and social care for which it responsible, obtain information about the needs of the person to whom that care is being or may be provided and the efficacy of that care.
133. In particular the Patient and Client Council, an organisation established by DHSSPS to represent the interests of the public and promote their involvement, is undertaking work in relation to patient empowerment issues in respect of rare diseases. It is supporting the development of the Northern Ireland Rare Disease Partnership and has recently undertaken a survey of the views of patients and carers of diagnosis of rare disease in Northern Ireland.
134. In Scotland there is a statutory duty on NHS Boards to promote public Involvement, enshrined in the National Health Services Reform (Scotland) Act 2004. The Scottish Government has also sponsored the development of ALISS (Accessing Local Information to Support Self-management), which aims to support people, including those with poor literacy skills, to find local support, much of which may be generic. For more information see <http://vimeo.com/23257731>

135. Welsh NHS bodies have a clear responsibility for continuous engagement with their local communities and to provide help and information as needed.
136. Patient organisations are also active in forming partnerships in Europe and globally to influence policy and foster research. The global solidarity of patient organisations is a useful counterweight to the local priorities which sometimes influence researchers and policy makers. Within Europe, patient organisations are strongly represented on the newly formed Committee of Experts on Rare Disease.
137. Patients and the public are represented at the highest levels of the NHS in England through membership of the Advisory Group for National Specialised Services. This advisory group advises Ministers on which specialised services are best commissioned nationally. The lay and patient members of this Advisory Group ensure that advice to ministers fully respects the unique perspective of patients and the public.
138. In Scotland, when the Patient Rights (Scotland) Act 2011 comes into force in 2012, patients will have the right that the healthcare they receive:
 - considers their needs;
 - considers what would be of optimum benefit to them;
 - encourages them to take part in decisions about their health and wellbeing, and provides information and support for them to do so.
139. This will ensure that Scottish patients can fully participate in decisions about their care and treatment. The Act also says that a patient's abilities, characteristics and circumstances should also be taken into consideration, allowing patients to have a real input into deciding how they are cared for and what treatment is best for them.
140. The expertise of those with rare conditions needs to be used, not only in day-to-day care, but also in the planning and organisation of services. Across the United Kingdom various approaches have been taken to enabling people with conditions, and their carers, to be involved in the design, development, implementation and evaluation of the services that are intended to support them.

141. Part of the Scottish Government's support for this approach has been the Voices programme, developed in collaboration with the British Heart Foundation and Chest Heart and Stroke Scotland, which has enabled patients to play a fuller role within various Managed Clinical Networks.
142. More locally most specialist teams providing highly specialised services are keen to seek the advice of the patients they serve. Patient experience surveys are a routine feature of NHS services: the results give useful information both on what is going well and where improvement can be made. Patient groups based around particular hospitals provide feedback in a more informal way on matters ranging from appointment systems to the decor for dedicated adolescent services.
143. From April 2013, for people living in England, Local HealthWatch and HealthWatch (England) will provide a platform for making the NHS and local government accountable to their local communities. They will enable the collective views of the people who use the health and social care services to influence national policy and to feedback on the services being provided locally. With so few people having each individual rare disease it will be very important that they are able to make their views known and give feedback on their care.

CONSULTATION QUESTION: In England, how best might this be facilitated with the introduction of Local HealthWatch and HealthWatch (England)?

8.1 Patient information and support

144. Information is key to patient empowerment.
145. Newly diagnosed patients and their families need information in a variety of languages and formats, and the information must be supplied at a pace that people struggling to come to terms with often devastating news can understand. Information prescriptions and information pathways set out information about different conditions, symptoms, diagnosis and treatments that patients can find helpful.
146. Information pathways provide information appropriate to the stage in a patient's journey. This supports a coherent approach from all those involved in the care plan. Information pathways have been developed and continue to be developed for long term conditions.

147. The Information Prescription System (IPS), hosted by NHS Choices, allows users both professional and public, to create information prescriptions (IPs) for long-term health needs. Information prescriptions provide people with and guide people to sources of reliable information from NHS Choices and its charity partners about their health and care – for example information about conditions and treatments, care services, benefits advice and support groups. IPs allow the user to tailor the amount of information provided at any one time around what the patient wants and can be used at different parts of an individual’s care pathway (e.g. diagnosis, treatment). Information prescriptions benefit patients, services users or carers by allowing them to feel more in control, and better able to manage their condition and maintain their independence, and can also help to reduce the anxiety, stress or confusion that many may feel in managing their condition.
148. Information is both widely and freely available on the internet but quality assurance is a problem. Internet search tools will reveal a huge number of sites offering information, and it is not possible to control or limit what happens on the internet. In England, the Department of Health in England supports The Information Standard, which is a certification scheme for health and social care information producers, to help the public identify trusted sources of health and social care information. The scheme has over 120 members with many more currently working towards membership. The scheme members include a number of NHS Trusts and leading national charities. The scheme awards a quality mark to its members and people can use this to identify trustworthy sources of information.
149. In addition, the “designated” centres providing services for rare conditions offer a range of written and internet information, which is reliable, authoritative and up-to-date. Patient organisations also provide helpful information on the diseases they represent and on NHS and other resources.
150. In England, the publication of the National Service Framework (NSF) for People with Long Term Conditions (2005) set out a number of key themes around empowerment, choice and control through personalised care planning and supported self care which could make a difference for people living with a rare condition. It also highlighted the different kinds of information available to support people with rare conditions including support networks and the role of the voluntary and community sectors to help people learn more about their condition. A number of workforce resources were also produced to support professionals. In addition the

- establishment of the Expert Patients Programme and the courses offered aim to help equip people with the confidence, skills and knowledge to manage their health condition successfully.
151. The generic model within the NSF puts a clear emphasis on ensuring care is integrated, the importance of team working (multi-disciplinary teams/networks) care co-ordination, improved health outcomes, engagement and empowerment. These are also crucial to improving the lives of people with rare conditions.
 152. Following a consultation exercise, it is intended that a policy framework to support people with long term conditions and their carers will be issued in Northern Ireland. This is intended to be a useful source of reference for people with long term conditions, setting out the services they and their carers should expect as a result of working in partnership with their Health & Social Care Trust and other care providers. Importantly it also outlines what they can do to help self manage their own conditions and ensure the best outcomes for their personal health and well-being and quality of life. The document is predicated on the belief that improving services and support for people with long term conditions cannot be taken forward by the Health & Social Care Trust and health professionals alone. It will involve working in partnership with people with long term conditions and their carers. It will also depend on building on the considerable amount of work already being undertaken with the voluntary and independent sectors, and with other Government Departments and agencies, to develop policies and joined up services to support people with long term conditions and their carers.
 153. Self management is at the very heart of the Scottish Government's work on long term conditions. The Self Management Strategy, published in August 2008, was informed by the lived experiences of people with long term conditions. Successful partnership working with many stakeholders has enabled the voluntary sector to link up with a range of initiatives and bring the Self Management Strategy to life. For example, the Self Management Fund aims to improve approaches to self management for people living with long term conditions. Mutually beneficial partnerships between patients, their families and those delivering healthcare services are delivering better, and more efficient services. To date, the Fund has supported people across Scotland to learn more about their long term conditions, and work in partnership with health and social care professionals to improve their quality of life.

154. Another strand of work that underpins self management work in Scotland is the ongoing development of ALISS (access to local information to support self management) which is helping to make people better managers of their own health by ensuring local information can be found easily online.
155. These examples show a continuing commitment by the UK to provide reliable patient information and support to those with rare conditions. Further information about the provision of high quality information will be set out in the forthcoming Information Strategy for England which is expected to be published by April 2012.

8.2 Orphanet

156. The Orphanet website at www.orpha.net is a major resource for patients and professionals. It is one of the most comprehensive websites on rare disease in the world and it is also one of the most reliable. The site provides not only scientific information for professionals but also details of where specialist centres and clinics for rare disease are located.
157. The United Kingdom participates in Orphanet through an information team based in Manchester.
158. This invaluable resource is part funded by the European Commission but each country must also make a contribution.
159. Since this site is fully accessible on the internet, there seems little point in trying to duplicate it in the United Kingdom. A sensible strategy is therefore to engage fully with this international effort.

CONSULTATION QUESTION: Do you agree that the United Kingdom should continue to participate in the Orphanet project?

8.3 NHS INFORM – Scotland

160. In August 2010, NHS Inform was launched in Scotland by the Cabinet Secretary for Health & Wellbeing. NHS Inform is available across three channels of delivery – online at www.nhsinform.co.uk, by telephone and face-to-face.
161. The online channel of delivery includes web based health information across a range of topics including; Health A-Z, Scottish Backs, Health in

my language, Health Rights. In addition, the support services directory provides information to communities regarding local support groups and services. The NHS in your area section offers opportunities for NHS Boards to supplement with their own health and service information. Common Health Questions and Behind the Headlines answer queries related to health and demystify health related news stories. On average, NHS Inform receives 36,500 web hits per month.

8.4 NHS Evidence

162. Alongside the Scottish website NHS Inform, the NHS Evidence website ensures that everyone working in health and social care has free access to the quality-assured, best-practice information required to inform evidence-based decision making, quickly and easily. On NHS Evidence users can search more than 150 sources simultaneously, including internationally respected evidence-based sources such as the Cochrane Library, British National Formulary and Map of Medicine. The types of resources available include guidelines, drug information, primary research and clinical summaries.

8.5 NHS Choices

163. NHS Choices is the UK's biggest health website. It provides a comprehensive health information service that puts the individual in control of their healthcare. The website helps people to make choices about their health from decisions about lifestyle to finding and using NHS services in England. It draws together the knowledge and expertise of:

- NHS Evidence;
- the Information Centre for Health and Social Care;
- the Care Quality Commission (CQC);
- many other health and social care organisations.

CONSULTATION QUESTION: What sources of patient information and support are available which are not listed in this plan?

8.6 Registers

164. A key tool in the struggle to understand rare disease is a register of patients suffering from the disease. Disease registers allow researchers to understand how the various symptoms develop or abate; how the disease progresses as people with a rare condition get older; and what heralds a

fatal outcome. A readily available list of patients makes it easier to recruit volunteers into trials of new therapies.

165. Registers are often regarded as the domain of researchers but people with a rare disease see value in being included on a register. In Scandinavia registers play a much greater role in the provision of quality care to patients. The Nordic Countries take advantage of their permanent residents having unique personal identification numbers to enable them to be able to perform register-based research and statistics. This number makes it possible to link information at the individual level from several registers.
166. There are National Patient registers in all the Nordic Countries, for both in-patient as well as out-patient care. International Classification of Disease 10 was introduced into hospital statistics across Scandinavia from the early 1990s and is also used by GPs.
167. Long term registers have been set up successfully by charities and patient organisations. One of the most successful is the Cystic Fibrosis register, which collects and validates information from patients throughout Europe. This register was originally established in Dundee but is now web-based. The register held by the MPS Society is one of the most comprehensive in the world for the mucopolysaccharide disorders. However, such registers require considerable resources.
168. Many of the long term registers of rare disease are run by pharmaceutical companies as a requirement of the drug license, but this disadvantages diseases for which no drug treatment is yet available. Where registers are held by pharmaceutical companies and more than one treatment becomes available for a rare disease our knowledge can become fragmented.
169. People with rare diseases are keen to see their information collected and analysed (subject of course to consent and statutory safeguards) so as to advance the understanding of their condition.
170. Another solution is to fund registers as part of the cost of care: service funding includes the cost of collecting relevant information. This is the principle which applies to hospital activity databases such as the Scottish Morbidity Record.

CONSULTATION QUESTION: Do you agree that registers are an important tool in rare disease and could be a core component of the service specification of an expert centre?

9 Sustainability

171. A plan must be sustainable. The actions proposed in this consultation document are all part of the normal business of the NHS or the health system in the four countries of the UK. In most cases the proposals will direct existing funding in a different way, or seek economies by better organisation and co-ordination.

10 Impact assessment and equality analysis

172. The consultation and the resulting UK Plan on Rare Diseases will not form part of any new or existing legislation and has been developed to be as cost neutral as possible. An Impact Assessment has not, therefore, been carried out. We hope that this consultation will help to identify whether there are any implications of reorganising existing resources to implement the suggested changes. We will use the information obtained through this consultation to inform whether an Impact Assessment is required before final publication of the UK Plan on Rare Diseases.
173. The general equality duty in the Equality Act (2010) requires public authorities, in the exercise of their functions, to have a due regard to the need to:
- eliminate unlawful discrimination, harassment and victimisation and other conduct prohibited by the Act;
 - advance equality of opportunity between people who share a protected characteristic and those who do not;
 - foster good relations between people who share a protected characteristic and those who do not.
174. As a result, the consultation document has been considered for its potential to impact on people who have the following protected characteristics: disability, sex, gender reassignment, race, age, sexual orientation, religion or belief, pregnancy and maternity. Overall, the consultation document and resulting UK Plan on Rare Diseases should have a positive impact on people with rare diseases, and often (as illustrated in the Equity Analysis) people with rare diseases fall into the protected characteristics identified by the Equality Act 2010. However, every opportunity to advance equality in the field of rare diseases should be taken to avoid creating new or exacerbating existing inequalities. For example, this could be done by improved recording and monitoring of protected characteristics in disease registers, in service delivery (and care co-ordination) and in access to medication/ health technologies.

CONSULTATION QUESTION: Are there any areas of work that the UK Plan on Rare Diseases needs to pay particular attention to in order to advance equality?

11 The consultation process

Criteria for consultation

This consultation follows the 'Government Code of Practice', in particular we aim to:

- formally consult at a stage where there is scope to influence the policy outcome;
- consult for at least 12 weeks with consideration given to longer timescales where feasible and sensible;
- be clear about the consultation's process in the consultation documents, what is being proposed, the scope to influence and the expected costs and benefits of the proposals;
- ensure the consultation exercise is designed to be accessible to, and clearly targeted at, those people it is intended to reach;
- keep the burden of consultation to a minimum to ensure consultations are effective and to obtain consultees' 'buy-in' to the process;
- analyse responses carefully and give clear feedback to participants following the consultation;
- ensure officials running consultations are guided in how to run an effective consultation exercise and share what they learn from the experience.

The full text of the code of practice is on the Better Regulation website at:

<http://www.bis.gov.uk/files/file47158.pdf>

Comments on the consultation process itself

If you have concerns or comments which you would like to make relating specifically to the consultation process itself please

Contact Consultations Coordinator
Department of Health
3E48, Quarry House
Leeds LS2 7UE

E-mail consultations.co-ordinator@dh.gsi.gov.uk

Please do not send consultation responses to this address.

Confidentiality of information

Information will be managed in accordance with the Information Charter or equivalent of the Country in which your response relates. The information we receive, including personal information, may be published or disclosed in accordance with the access to information regimes (primarily the Freedom of Information Act 2000 (FOIA) and equivalent legislation in Scotland, Wales and Northern Ireland, the Data Protection Act 1998 (DPA) and the Environmental Information Regulations 2004).

If you want the information that you provide to be treated as confidential, please be aware that, under the FOIA, there is a statutory Code of Practice with which public authorities must comply and which deals, amongst other things, with obligations of confidence. In view of this, it would be helpful if you could explain to us why you regard the information you have provided as confidential. If we receive a request for disclosure of the information we will take full account of your explanation, but we cannot give an assurance that confidentiality can be maintained in all circumstances. An automatic confidentiality disclaimer generated by your IT system will not, of itself, be regarded as binding on the Department of Health, England.

The Department of Health in England will process your personal data in accordance with the DPA and, in most circumstances, this will mean that your personal data will not be disclosed to third parties.

Summary of the consultation

A summary of the response to this consultation will be made available before or alongside any further action, such as laying legislation before Parliament, and will be placed on the Department of Health, England's Consultations website at <http://www.dh.gov.uk/en/Consultations/Responsestoconsultations/index.htm>

How to respond

This consultation starts on 29 February 2012. Responses should be sent using the attached consultation response form no later than 25 May 2012 to:

rarediseasesconsultationresponses@dh.gsi.gov.uk

Alternatively, copies can be sent by post to:

Sarah Bramley-Harker
Department of Health
Screening & Specialised Services Team
Room 5W35, Quarry House
Quarry Hill
Leeds LS2 7UE.

12 Consultation response form

UK Plan on Rare Diseases

Closing date for responses: Friday 25th May 2012. Please send your replies to: rarediseasesconsultationresponses@dh.gsi.gov.uk or alternatively send by post to: Sarah Bramley-Harker
Department of Health
Screening & Specialised Services Team
Room 5W35, Quarry House
Quarry Hill
Leeds, LS2 7UE.

Please fill in and/or tick the appropriate response.

Name
Contact address
Organisation representing (if appropriate)
Postcode
Contact telephone
Email

Before submitting your response please make sure that it has been saved in a name [e.g. A N Other] that will make it easier for us to track. Many thanks.

Freedom of Information

This consultation document has been produced by the four UK Health Departments. The information you provide in response to this consultation will be managed in accordance with the **Information Charter, or equivalent**, of each UK Health Department.

Information provided in response to this consultation, including personal information, may be published or disclosed in accordance with the access to information regimes. The relevant legislation in this context is the Freedom of Information Act 2000 (FOIA) and the Data Protection Act 1998 (DPA) and equivalent legislation in Scotland, Wales and Northern Ireland.

If you want the information that you provide to be treated as confidential, please be aware that, under the FOIA, there is a statutory Code of Practice with which public authorities must comply and which deals amongst other things, with obligations of confidence. In view of this, it would be helpful if you could explain to us why you regard the information you have provided as confidential. If we receive a request for disclosure of the information we will take full account of your explanation, but we cannot give an assurance that confidentiality can be maintained in all circumstances. An automatic confidentiality disclaimer generated by your IT system will not, of itself, be regarded as binding.

Your personal data will be processed in accordance with the DPA and in most circumstances this will mean that your personal data will not be disclosed to third parties. However, the information you send may need to be passed on to colleagues within the other UK Health Departments and/or published in a summary of responses to this consultation.

I do not wish my response to be passed to other UK Health Departments

I do not wish my response to be published in a summary of responses

Please indicate the country the consultation and your comments relate:

<i>UK-wide</i>	<input type="checkbox"/>	<i>and/or:</i>	
<i>England</i>	<input type="checkbox"/>	<i>Northern Ireland</i>	<input type="checkbox"/>
<i>Scotland</i>	<input type="checkbox"/>	<i>Wales</i>	<input type="checkbox"/>

Are you responding: – *as a member of the public*

– *as a health or social care professional*

– *on behalf of an organisation*

Area of work:

NHS	
Social Care	
Private Health	
Third Sector	
Regulatory Body	
Professional Body	
Education	
Trade Union	
Local Authority	
Trade Body	
Other (Please give details)	
Independent Contractor to NHS	
Manufacturer	
Supplier	
Other (where relevant)	

In which of the following areas do you live: (please tick one box only)	
North East	
North West	
West Midlands	
South East	
London	
Humberside/Yorkshire	
East Midlands	
East of England	
South West	
No answer	
Scotland	
Wales	
Northern Ireland	

If you are responding on behalf of an organisation, please indicate which type of organisation you represent:

NHS	
Social Care	
Private Health/Independent Sector	
Third Sector	
Regulatory Body	
Professional Body	
Education	
Trade Union	
Local Authority	
Trade Body	
Other (Please give details)	

1 What is your sex?*

Tick one box only.

Male	<input type="checkbox"/>
Female	<input type="checkbox"/>
Prefer not to say	<input type="checkbox"/>

2 What is your Age?*

Age	
Prefer not to say	

3 Are your day to day activities limited because of any health problem or disability which has lasted, or is expected to last at least 12 months?

Tick one box only.

Yes, limited	<input type="checkbox"/>
Yes, limited, a little	<input type="checkbox"/>
No	<input type="checkbox"/>
Prefer not to say	<input type="checkbox"/>

4 Do you look after, or give any help or support to family members, friends, neighbours or others because of either long term physical or mental ill-health/disability or problems related to old age?

Tick one box only.

Yes	
No	
Prefer not to say	

5 What is your ethnic group?

Tick one box only.

A White

British

Irish

Any other White background, write below

B Mixed

White and Black Caribbean

White and Black African

White and Asian

Any other Mixed background, write below

C Asian, or Asian British

Indian

Pakistani

Bangladeshi

Any other Asian background, write below

D Black, or Black British

Caribbean

African

Any other Black background, write below

E Chinese, or other ethnic group

Chinese

Any other, write below

F Prefer not to say

6 What is your religion or belief?

Tick one box only.

Christian includes Church of Wales, Catholic, Protestant and all other Christian denominations.

None

Christian

Buddhist

Hindu

Jewish

Muslim

Sikh

Prefer not to say

Other, write below

7 Which of the following best describes your sexual orientation?

Tick one box only.

Only answer this question if you are aged 16 years or over.

Heterosexual Straight

Lesbian/Gay Woman

Gay Man

Bisexual

Prefer not to say

Other, write below

Consultation Questions

1. CONSULTATION QUESTION: Do you agree that commissioners of services should explore the potential of expert clinical systems to reduce diagnostic delay, particularly in neurology and genetics?

Comments

2. CONSULTATION QUESTION: Can you suggest ways of rare disease featuring more prominently in speciality training for doctors?

Comments

3. CONSULTATION QUESTION Do you agree that the UK National Screening Committee should take into account the benefit of screening in reducing the 'diagnostic odyssey' and in allowing informed choice for subsequent family planning?

Comments

4. CONSULTATION QUESTION How can the NHS best ensure research in rare diseases carried out by the NIHR biomedical research centres and units is rapidly transferred into practice for the benefit of UK patients and their families and carers?

Comments

5. CONSULTATION QUESTION: Do you agree that commissioners of care for people with rare diseases should assess options for improved care coordination, including named care coordinators?

Comments

6. CONSULTATION QUESTION: Do you agree that this list of criteria for expert centres should be the basis for future shaping of services:
- co-ordinated care;
 - adequate caseload for expertise;
 - not dependent on a single clinician;
 - arrangements for the transition from children's to adults' services;
 - engaged with people with rare conditions;
 - research active?

Comments

7. CONSULTATION QUESTION: Do you agree that each expert centre must know its network of local hospitals, and the local hospitals must know the pathway to the expert centre which will offer help, support, advice and assistance?

Comments

8. CONSULTATION QUESTION: In England, how best might this be facilitated with the introduction of Local HealthWatch and HealthWatch (England)?

Comments

9. CONSULTATION QUESTION: Do you agree that the United Kingdom should continue to participate in the Orphanet project?

Comments

10. CONSULTATION QUESTION: What sources of patient information and support are available which are not listed in this plan?

Comments

11. CONSULTATION QUESTION: Do you agree that registers are an important tool in rare disease and could be a core component of the service specification of an expert centre?

Comments

12. CONSULTATION QUESTION: Are there any areas of work that the UK Plan on Rare Diseases needs to pay particular attention to in order to advance equality?

Comments

Before submitting your response, please make sure that it has been saved in a name [e.g. A N Other] which will make it easier for us to track.

Please email or send this form back by 25 May 2012 to:

by Email: rarediseasesconsultationresponses@dh.gsi.gov.uk

**by Post: Sarah Bramley-Harker
Department of Health
Screening & Specialised Services Team
Room 5W35, Quarry House
Quarry Hill
Leeds, LS2 7UE.**

Many thanks for your response.



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