



RARE DISEASE | UK

# Improving Lives, Optimising Resources:

## A Vision for the UK Rare Disease Strategy



The National Alliance for people with  
rare diseases & all who support them

[www.raredisease.org.uk](http://www.raredisease.org.uk)



# About Rare Disease UK

Rare Disease UK (RDUK) is the national alliance for people with rare diseases and all who support them. Our membership is open to all and includes patient organisations, clinicians, researchers, academics, industry and individuals with an interest in rare diseases.

RDUK was established by Genetic Alliance UK, the national charity of over 130 patient organisations supporting all those affected by genetic conditions, in conjunction with other key stakeholders in November 2008 following the European Commission's Communication on Rare Diseases: Europe's Challenges.

Subsequently RDUK successfully campaigned for the adoption of the Council of the European Union's Recommendation on an action in the field of rare diseases. The Recommendation was adopted unanimously by each Member State of the EU (including the UK) in June 2009. The Recommendation calls on Member States to adopt plans or strategies for rare diseases by 2013.

RDUK is supported by an unrestricted educational grant from the ABPI Orphan Diseases Industry Group and the Orphan Disease Industry Group Partnership. A list of these companies is available at [www.raredisease.org.uk/members.htm](http://www.raredisease.org.uk/members.htm)

RDUK is campaigning for a strategy for integrated service delivery for rare diseases. This would coordinate:

- Research
- Prevention, diagnosis and screening
- Treatment
- Care and support
- Information
- Commissioning and planning

into one cohesive strategy for all patients affected by rare disease in the UK. As well as securing better outcomes for patients, a strategy would enable the most effective use of NHS resources.



## Acknowledgements

We wish to express our gratitude to the hundreds of individuals and organisations that contributed to the production of this report. Without this expertise and insight, offered from a broad array of backgrounds, the production of this report would not have been possible.

We would particularly like to thank the chairs and members of the Working Groups for volunteering their time and for their support in guiding and shaping the development of this report.

### The Rare Disease UK team

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# Chair's Foreword

Ever since its establishment, the fundamental principle underpinning the National Health Service has been that patients would be treated according to their needs and that treatment would be free at the point of delivery irrespective of the individual's ability to pay.

When the NHS was founded most rare diseases were not diagnosable, much less treatable in any meaningful way. Thanks to scientific advances - many of them arising from the genetics revolution of the last twenty years - most rare diseases can now be accurately diagnosed and whilst most remain untreatable, a small but growing number of rare conditions are able to benefit from innovative therapies developed as a result of cutting edge research and development. A significant proportion of this has been undertaken by universities and companies based in the UK.

However, as the "Experiences of Rare Diseases: An Insight from Patients and Families"<sup>1</sup> report published by Rare Disease UK in December 2010 shows a significant proportion of those with rare diseases do not receive an adequate response to their needs from the NHS. The numbers are not small. It is estimated that over 3.5 million people will be affected by a rare disease in the UK at some point in their lives. If 46% have to wait more than a year to get a diagnosis that represents at least 1.6 million person years of unmet need and potentially avoidable harm. Considering diagnosis takes over 5 years for 20% of these patients and over 10 years for 12%, this figure is likely to be even greater. Unfortunately, in our experience patients' needs are often not adequately met even after diagnosis.

It is not that those who work for the NHS are uncaring or lack the will to respond to the needs of patients and families with rare diseases. Rather it is that the systems and structures are not necessarily in place to respond to the needs of many of those affected. Of course it is not all bad; the NHS has examples of world class services for patients and families with rare diseases, and the report highlights just some of them. But what is lacking is a coherent strategy that will bring together the existing knowledge and expertise, wherever it is located (in the NHS, in our universities, in industry, in patient groups and medical research charities) in a coherent, logical and equitable way so that the founding principle of the NHS can become a reality for all patients with rare diseases in the UK.

The Government committed the UK to develop such a strategy when it signed the Council of the European Union's Recommendation in June 2009<sup>2</sup>. Since then, Rare Disease UK has been working with all interested parties, including those in the health departments and the NHS in all four home nations of the UK, to develop a comprehensive strategy.

Our recommendations for a strategy are presented in this report. It is practical, realistic and attainable in its proposals. Many of the actions proposed can be taken now with little alteration of existing practices. Others will take longer but with a sustained commitment to action and a willingness to be bold there is no reason why they too cannot be achieved sooner rather than later.

Rare Disease UK is grateful to all those who willingly gave their time and expertise to bring this report into being. We look forward to a continued collaboration with the health departments, the NHS and all other interested parties in moving forward to the implementation of the proposals contained herein and the realisation of the entirely legitimate aspirations of patients and families with rare diseases for an effective, timely and appropriate response to their needs by the NHS wherever they happen to live in the UK.

Working together we can secure the best use of scarce expertise and resources, maximise the health gain for all those with rare conditions and create a framework for the research necessary to support the development of innovative therapies for unmet medical needs. This makes sound sense economically, clinically, scientifically and for all those in need of high quality care and support.



Alastair Kent OBE  
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1. Limb et al, "Experiences of Rare Diseases: An insight from Patients and Families" (December 2010) available at: <http://www.raredisease.org.uk/documents/RDUK-Family-Report.pdf>
2. Recommendation on an action in the field of rare diseases (2009/C 151/02).



# Introduction

The adoption of the Council of the European Union's Recommendation on an action in the field of rare diseases in June 2009 provided a window of opportunity for the UK. The main requirement of the Recommendation is the development and implementation of a plan or strategy for rare diseases by 2013. Rare Disease UK (RDUK) was eager for this opportunity to be capitalised on; an effective strategy for rare diseases would not only improve the quality of services and health outcomes for people affected by rare diseases and their families, but would also ensure a more efficient use of NHS resources.

A rare disease is defined by the EU as affecting fewer than 5 in 10,000 of the general population.<sup>3</sup> Although there are no precise figures on the amount of people affected by rare diseases in the UK due to a failure to collect this data, best estimates arising from the Council Recommendation suggest that 1 in 17 people will be affected by a rare disease at some point in their lives. This amounts to 3.5 million people across the UK.

## **Collectively, rare diseases are not rare and they represent a significant health burden to the health services in the UK.**

Patients with rare diseases already make heavy demands on the resources of both health and social services, but these resources are often used inefficiently due to delays in diagnosis, misdiagnosis, fragmented care, a lack of information, few guidelines on the effective management of conditions and limited effective treatment options.

It is not all bad, however; there are many examples of excellent practice throughout the UK and we highlight just a few of these in this report. These examples demonstrate that it is possible to provide high quality services and support to people with rare diseases. Many of these services have been shown to save money or are cost-neutral by delivering improved and more efficient care, leading to better health outcomes. Unfortunately, most patients with rare diseases cannot access such high quality services and those services in existence have generally developed ad hoc with little consideration of the overarching needs of patients with rare diseases. We believe that with a strategic approach to the development of services for rare diseases, we can move towards the situation where these services become the norm as opposed to the exceptions. The UK has the opportunity to lead in terms of research, the development of treatments and care guidelines and in the provision of high quality, innovative services for patients with rare diseases.

RDUK was eager to work with our members and the broad stakeholder community to develop this report to demonstrate how patients' and families' lives can be improved, and NHS resources optimised, by taking some creative approaches and by building on the innovative ways of service planning and delivery that are already proving successful in the UK. (Information about how we compiled this report is available in the Methodology section.)

Many of the recommendations outlined in this report can be implemented easily and swiftly. Others may be more difficult or will take longer; however, this should not be an excuse to avoid addressing these issues. A strategy for rare diseases should outline a clear course of action accompanied by timescales for implementation. There may of course be other solutions, or there may be other bodies that can best carry out the actions we propose, and we are open to, and welcome, further ideas and discussion.

For too long, patients with rare diseases have had to face inequitable access to high quality services, treatment and support. Now is the time for health departments across the UK to demonstrate the commitment to bringing an end to this inequity by addressing the situation in a well-planned, strategic approach.

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3. Council Recommendation on an action in the field of rare diseases, June 2009

# Overarching principles for a strategy for rare diseases:

## Collaboration

A strategy for rare diseases should be developed in partnership between the health departments of all four home nations. Joint working is required due to the small numbers of patients affected by each rare disease, especially those at the very rare end of the spectrum. No one nation can solve the needs of all rare disease patients alone. A strategy for rare diseases should also facilitate collaboration across Europe and internationally where appropriate.

A collaborative approach should be taken to the development and implementation of a strategy. Key stakeholder groups, including patients and patient organisations, are a vital source of information and experience, and processes should be put in place to engage systematically with these groups in a partnership to achieve this goal.

## Oversight and accountability

The oversight and delivery of a strategy for rare disease must be included within the remit of a designated team/unit in each of the UK's health departments.

The former Chief Medical Officer for England, Sir Liam Donaldson, included a series of recommendations in a chapter entitled 'Rare is Common' in his 2009 Annual Report. One of these recommendations was for a National Clinical Director for Rare Diseases to 'oversee the development of clear standards and pathways for the treatment and surveillance of rare diseases, with national registers to support service planning and delivery as well as research.'<sup>4</sup> We fully support this recommendation and believe that this position should command sufficient stature to achieve the stated aims. Similar national clinical leads should also be designated in Scotland, Wales and Northern Ireland.

## Innovation

Effective planning and service provision for rare diseases necessitate innovative ways of working; a strategy for rare diseases should encourage and facilitate this. Many of the recommendations outlined in this report can be achieved through greater collaboration between the NHS and patient organisations or through public/private partnerships, for instance.

## Outcomes

The outcomes and effectiveness of a strategy for rare diseases should be monitored. This will require health departments to develop clear ways of assessing and measuring the impact of a strategy.

4. '2009 Annual Report of the Chief Medical Officer', Department of Health, March 2010

# Methodology

RDUK was established by Genetic Alliance UK in November 2008 in recognition of the need for a wide range of stakeholders to be involved in the development of an effective strategy for rare diseases. One of the benefits of establishing RDUK as a multi-stakeholder organisation is that we have been able to gather evidence widely and tap into the expertise in all of these stakeholder groups. We have always aimed to take a collaborative approach towards an ultimate common goal: to make the most effective use of NHS resources so as to provide the best possible outcome for patients and families affected by rare diseases.

RDUK is also able to access a vast knowledge base that has been accumulated over the past 20 years of Genetic Alliance UK's existence. This knowledge base continues to grow daily through our interactions with patients, carers, families and patient organisations.

Alongside desk research and an extensive number of meetings and interviews with a broad range of stakeholders, in order to develop this report we have undertaken a number of activities to gather evidence systematically. These activities are listed below.

## Working Groups

Central to the process of developing this report were five multi-stakeholder Working Groups established by RDUK to focus on the following areas:

- Coordination of Research
- Prevention and Diagnosis
- Commissioning and Planning
- Patient Care, Information and Support
- Delivering Coordinated Care

The Working Groups included representatives from, but not limited to:

- Patients/family members
- Representatives from patient organisations
- Researchers
- Clinicians
- Healthcare professionals
- Geneticists
- Representatives from the pharmaceutical industry
- Commissioners of specialised services

The list of members of each Working Group is available in Annex 1.



Each of the groups was free to decide its own lines of inquiry; RDUK did, however, provide suggested terms of reference to each of the groups, which they subsequently adapted to suit the needs of the group. We also asked the Working Groups to consider the following principles in developing recommendations:

- Recommendations should be responsive to changing patterns of NHS organisation.
- Recommendations should be relevant to all four home nations and adaptable to the different NHS structures in each.
- In recognition of the current financial climate, recommendations should lead to a more efficient use of resources to maximise the health gain for patients with rare diseases, or where appropriate, the targeted use of resources for better outcomes.
- To gather as many examples of good practice as possible, both to ensure that current good practice is being optimally utilised and in order to provide models for new services.

The Working Groups met over a period of a year. The approach of each Working Group was tailored to its particular needs; despite this the structure of the inquiry followed a broadly similar pattern:

1. An assessment of the current situation to identify needs
2. The development of initial solutions to problem areas
3. Collection of good practice to act as models of service provision
4. Framing concrete recommendations on the basis of the evidence gathered

## Consultation

A consultation document was produced outlining the initial recommendations of the five Working Groups. Aimed at stimulating further input, the consultation offered an overview of the discussions of the Working Groups. We encouraged respondents to comment on the recommendations and issues identified, to elaborate on the recommendations with examples of good practice, to highlight problem areas and to suggest other solutions.

The consultation was distributed to RDUK members and to other key stakeholders with a two month period over September and October 2010 in which to respond. We received a total of 92 written responses from the following groups:

- 34 patient organisations
- 6 pharmaceutical/biotech companies and 1 umbrella body
- 17 patients or a family member
- 11 clinicians and healthcare professionals
- 10 professional bodies/networks
- 9 researchers/academics
- 2 public bodies
- 3 others

The full list of those who responded to the consultation can be seen in Annex 2.

All responses received were used to inform the development of our final recommendations.

## Focus Groups in Devolved Nations

Although there were members of the Working Groups professionally based across all four of the UK's home nations, we were aware that there was not a representative from each of the devolved nations at each meeting. This could have led to a failure to take into account the increasingly divergent nature of the NHS and of health policy in each of the home nations when framing the recommendations. It was crucial not to overlook these differences.

In order to address the issue, we held three focus groups in Scotland, Wales and Northern Ireland respectively during November 2010. The focus groups broadly reviewed the recommendations highlighted in the consultation document, highlighting specific considerations or differences where appropriate.

The focus groups comprised a range of stakeholders similar to the composition of the Working Groups as well as representation from the health departments of the devolved administrations. A full list of focus group attendees is available in Annex 3.

The most striking outcome of the focus groups is that, while there are differences in the structures and organisations between the home nations and consideration should be given to aspects such as geography, the needs of patients with rare diseases generally remain the same and the principles underlying our recommendations are applicable across the UK.

RDUK's Management Committee includes representatives from Scotland, Wales and Northern Ireland who have also helped to ensure that recommendations are appropriate to their respective nations.

## Survey of patients' and families' experiences

Over the summer of 2010, RDUK conducted a survey of patients' and families' experiences of rare diseases. This wide-ranging survey dealt with topics including access to information and support, coordination of care, access to treatment, diagnosis, and participation in research. The aim of the survey was to gain a better understanding of the issues faced by patients and families affected by rare diseases in accessing services and support, but also to highlight areas of good practice.

We received 600 responses from patients and families affected by 119 different rare conditions. To obtain responses, the electronic survey was distributed to members of RDUK, many of which circulated it to their own contacts. Hard copies were available on request. Responses to the survey were split almost evenly between individuals with a rare condition (47%) and family members/carers of people with a rare condition (49%). The remaining 4% were from other interested parties, including health professionals responding on behalf of their patient. Responses to the survey were also broadly in proportion to the population of the home nations of the UK with 78% living in England, 12% in Scotland, 6% in Wales and 3% in Northern Ireland. The remaining 1% were from patients living in the Channel Islands.

The findings were published in a report 'Experiences of Rare Diseases: An Insight from Patients and Families' (available at: <http://www.rare-disease.org.uk/documents/RDUK-Family-Report.pdf>).

We refer to the findings where appropriate throughout this report.

## EUROPLAN Conference

The European Project for Rare Diseases National Plans Development (EUROPLAN) is a three-year project of the European Commission's Programme of Community action in the field of Public Health (2003 - 2008), which began in April 2008. The National Specialised Commissioning Group is an associated partner in the project. The ultimate aims of the project are to create guidance to aid the development of national plans/strategies for rare diseases, and to establish indicators to evaluate the impact of these plan/strategies.

One of the work streams of the project involved organising conferences in 15 Member States to gather more information on the provision of services for rare diseases at a national level to inform the Commission in developing their final recommendations. RDUK was responsible for hosting the UK conference on the 16th November 2010.

With over 80 attendees, including representation from the health departments, the conference provided an opportunity to gather more valuable evidence to inform the recommendations outlined in this report. The full report of the EUROPLAN conference is available on the RDUK website.

A list of conference attendees is available in Annex 4.

## Joint AMRC/RDUK workshop on research into rare disorders

On the 9th December 2010, RDUK held a one-day joint workshop with the Association of Medical Research Charities (AMRC) to look at funding and conducting research into rare diseases. The workshop provided an opportunity for patient organisations and industry to convene to discuss some of the issues that they have in relation to research, and to discuss possible solutions. The outcomes of the day were captured to help inform the Coordination of Research section of this report.

A full list of attendees is available in Annex 5.

## Management Committee

We have had input from members of the multi-stakeholder RDUK Management Committee throughout the process of developing this report.

A list of the Management Committee members can be found in Annex 6.

# Summary of the recommendations

## Coordination of Research

Current situation	Recommendations	Target of recommendation
There is limited collaboration between researchers working on rare diseases.	1. Mechanisms should be put in place to improve collaboration between rare disease researchers.	a. Development of (and funding for) clinical research networks focused in rare disorders. NIHR.
		b. Established research networks should support research into rare diseases. NIHR Clinical Research Networks and others.
		c. Existing networks should help inform the development of new networks. NIHR Clinical Genetics Network and others.
There is limited funding provision for research into rare diseases.	2. Funding bodies should be encouraged to support research into rare diseases.	a. Inclusion of rare disease experts on funding boards' committees. MRC, NIHR and other funding bodies.
		b. Raise awareness of research supported by patient organisations. UK branch of Orphanet and others.
		c. UK participation in E-Rare. MRC.
There is a lack of basic epidemiological and clinical data on rare diseases.	3. Support should be given to develop and sustain systems for data collection and disease registries for patients with rare diseases.	a. Development of disease registries. NHS, patient organisations, funding bodies, industry, researchers, clinicians.
		b. Sustainable financial support to registries. Funding bodies.
		c. Increased collaboration and harmonisation of data sets. Symposium of funders.
		d. Ensure long-term funding for surveillance units. Health departments.
		e. Implementation of the International Classification of Diseases 11 (ICD 11). NHS.
		f. Development and support of databases on disease genotype/phenotype correlations. NHS, CRNs, funding bodies e.g. NIHR, MRC, Wellcome Trust.

Current situation	Recommendations	Target of recommendation
<p>There are difficulties in the development of new diagnostic tests for rare diseases.</p>	<p>4. Measures should be taken to encourage the development and approval of diagnostic tests.</p>	<p>a. Implementation of a coordinated national strategy for the development of rare genetic tests. NHS, UKGTN, BSHG.</p>
		<p>b. Expansion of approved genetic tests and its translation into laboratory funding. UKGTN, regional Genetics Laboratories, NHS Commissioning Board/Commissioners.</p>
		<p>c. Coordinated approaches and specific funding for laboratory developmental work. NIHR, MRC.</p>
		<p>d. New DDD project to offer the opportunity to use new breakthrough technique. NIHR.</p>
<p>The need to obtain Research and Development (R&amp;D) approval from numerous sites slows and inhibits rare disease research.</p>	<p>5. The system for gaining R&amp;D approval for research that spans the UK should be streamlined.</p>	<p>a. Reappraisal of local R&amp;D approval system to speed up the overall process. Health departments.</p>
		<p>b. R&amp;D approval to cover all four nations. Health departments.</p>
		<p>c. Rare disease R&amp;D approval to be proportionate to the complexity of the project. Human Genomic Strategy Group, NIHR Clinical Genetics Networks, NRES.</p>
		<p>d. Standardisation of R&amp;D fees across UK, and according to project complexity. Health departments.</p>
<p>There are difficulties in therapeutic and prevention research, including research methodologies applicable to rare disorders and the development of orphan drugs for children.</p>	<p>6. Explore and promote research and debate into appropriate and acceptable research methods into the prevention and treatment of rare disorders.</p>	<p>Funding bodies, MHRA, NRES, Health Research Agency (as proposed by the Academy of Medical Sciences).</p>
<p>There is currently very little research on the management of rare disease patients.</p>	<p>7. Commission research on health service delivery for patients with rare disorders and promote and support the development of guidelines as tools to improve care management.</p>	<p>Funding bodies, including the NIHR.</p>

# Prevention and Diagnosis

Current situation	Recommendations		Target of recommendation
<p>There is a lack of awareness and identification of rare diseases amongst healthcare professionals, often resulting in a delay in diagnosis or misdiagnosis of rare disease patients.</p>	<p>1. Increase healthcare professionals' knowledge and awareness of rare diseases.</p>	<p>a. Inclusion of a rare disorders module in medical curriculum.</p>	<p>Royal Colleges and Deans of medical schools in the UK.</p>
		<p>b. Ongoing CPD/CME training.</p>	<p>Royal Colleges and Regional Deaneries.</p>
		<p>c. Secondary medical care providers training in basic genetics.</p>	<p>Health departments through initiatives such as National Genetics Education and Development Centres.</p>
		<p>d. Development of e-learning packages to increase overall awareness of rare diseases by qualified professionals.</p>	<p>National Genetics Education and Development Centre, in partnership with the Royal Colleges under the guidance of the Joint Committee on Medical Genetics.</p>
		<p>e. Formal recognition of RGS teams' educational role.</p>	<p>NHS.</p>
	<p>2. There is the need for improved linkage between specialist centres and local services to enable education of local healthcare professionals.</p>	<p>a. Staff exchanges to ensure information update.</p>	<p>Clinical service managers, healthcare professionals.</p>
		<p>b. Formal recognition of specialist centres teams' educational role.</p>	<p>NHS, patient organisations.</p>
		<p>c. 'Hub and spoke' model between specialised and local services.</p>	<p>Commissioners.</p>



Current situation	Recommendations	Target of recommendation
<p>It can be difficult for both professionals and patients to access reliable, up-to-date information on rare diseases and who the specialist(s) is/are in a particular condition.</p>	<p>3. Improve access to reliable information on rare diseases to make it easier for the public and professionals to obtain information.</p>	<p>a. UK online portal linking to reliable sources of information and guidelines. All UK health departments.</p>
		<p>b. Appraisal of healthcare professionals' information needs and gaps in existing resources. Joint Committee on Medical Genetics, Royal Colleges and Deans of medical schools in the UK.</p>
		<p>c. Ensure adequate Orphanet matched funding. Department of Health.</p>
		<p>d. Named responsible clinician against each condition in the NHS Directory of Genetic Testing. UKGTN.</p>
		<p>e. Ensuring training on the use of diagnostic tools are included as part of projects. Funding bodies.</p>
		<p>f. Development of an "index of suspicion" to better guide doctors when referring to specialists. Joint Committee on Medical Genetics.</p>
<p>The criteria currently used by the National Screening Committee (NSC) to determine suitability for newborn screening tend to militate against rare conditions.</p>	<p>4. Appropriate rare diseases need to be considered for inclusion in the newborn screening programme.</p>	<p>a. Re-appraisal of current criteria in screening for rare conditions. National Screening Committee.</p>
		<p>b. Inclusion in the NSC of representatives with experience in rare conditions. National Screening Committee.</p>
		<p>c. Consider recommendations by EU Newborn Screening Study (launch June 2011). National Screening Committee.</p>
		<p>d. Cost effective public-private partnership. National Screening Committee.</p>

Current situation	Recommendations		Target of recommendation
There is inequity of access to diagnostic tests across the UK.	5. There must be improved access to diagnostic tests in areas of the UK where this is lacking to ensure equity of access throughout the country.	a. Review of the architecture of testing for rare conditions in the UK.	NHS Commissioning Board in collaboration with the NSD, WHSSC and the HSCB.
		b. Reduce unnecessary testing by developing guidelines, improving resources and ensuring expert guidance.	UKGTN.
	6. Access to carrier tests for individuals and groups considered to be at significant risk of a specific condition should be facilitated and promoted.	a. Introduce testing programmes for at-risk groups.	Commissioners, UKGTN, professional societies e.g. BSHG.
		b. Carrier tests should take into account the frequency of the particular condition within the population, and should be available to those that are considered high risk.	Commissioners, UKGTN, professional societies e.g. BSHG.
		c. Ensure long-term sustainability of carrier testing programmes in existence.	Commissioners, UKGTN, professional societies e.g. BSHG.

# Commissioning and Planning

Current situation	Recommendations		Target of recommendation
<p>There is a wide variation in the health services available for patients with rare diseases across the UK.</p>	<p>1. Commissioning and planning systems for rare diseases should ensure equitable access to services and treatments across the UK, regardless of a patient's location.</p>	<p>a. Ensure commissioning and planning of services is carried out at the appropriate population level.</p>	<p>Health departments, NHS Commissioning Board, NSD, WHSSC, HSCB, local commissioners/planners.</p>
		<p>b. Review current services and policies regarding access across home nations.</p>	<p>NHS Commissioning Board, NSD, WHSSC, HSCB.</p>
		<p>c. Coordination among UK's specialised commissioning bodies to improve access to services.</p>	<p>NHS Commissioning Board, NSD, WHSSC, HSCB.</p>
<p>There is often a lack of coordination between what is commissioned or planned centrally and what is commissioned or planned at the local level.</p>	<p>2. Commissioning or planning structures should facilitate the coordination of what is commissioned or planned centrally and what is commissioned or planned at a local level.</p>	<p>a. Strong oversight body to ensure this integration and linkage.</p>	<p>NHS Commissioning Board, NSD, WHSSC, HSCB.</p>
		<p>b. Develop and implement guidelines to ensure integration of services.</p>	<p>Specialised commissioning/planning bodies in partnership with local commissioners and planners, clinicians and patient organisations.</p>
		<p>c. Taking into account that the distribution of patients with certain conditions will not be even.</p>	<p>All commissioning/planning bodies.</p>
<p>Provision of funding for specialised services for patients with rare diseases is perceived by some to be diverting resources away from local services.</p>	<p>3. The value of specialised services needs to be recognised and there should be resources safeguarded to fund these services.</p>	<p>a. Protected, flexible budgets should be allocated to the specialised commissioning/planning body.</p>	<p>Health departments.</p>

Current situation	Recommendations	Target of recommendation	
<p>Specialised services do not exist for every rare condition, and this can result in inequitable levels of care depending on which condition a patient has.</p>	<p>4. Structures should be in place to ensure patients are able to access the best care and support regardless of whether a specialised service exists for that condition.</p>	<p>a. Specialised services developed for clusters of conditions with similar needs. (See also: Delivering Coordinated Care – Recommendation 1a)</p>	<p>NHS Commissioning Board, NSD, WHSSC, HSCB.</p>
		<p>b. Appraisal of which conditions need to be prioritised for specialised services.</p>	<p>NHS Commissioning Board, NSD, WHSSC, HSCB.</p>
		<p>c. Sharing of good practices among services.</p>	<p>All commissioning/ planning bodies, those delivering the service.</p>
		<p>d. Support patient organisations and clinicians to apply for specialised services.</p>	<p>NHS Commissioning Board, NSD, WHSSC, HSCB.</p>
		<p>e. Periodical review of services to assess utility.</p>	<p>NHS Commissioning Board, NSD, WHSSC, HSCB.</p>
<p>UK patients with rare diseases are being denied access to orphan medicines that have been granted European marketing authorisation.</p>	<p>5. There is an urgent need to reassess the mechanism and methodology by which the value of medicines for rare conditions is appraised for reimbursement on the NHS, to ensure improved and equitable access to licensed medicines from which patients will benefit.</p>	<p>a. Evaluation methods and processes should be refined for orphan medicines.</p>	<p>NICE, SMC, AWMSG, in collaboration with others including NIHR HTA and patient organisations.</p>
		<p>b. Funding for specialised drugs/treatments/ interventions for rare diseases should be organised nationally from a central source.</p>	<p>NHS.</p>
		<p>c. New VBP to consider issues specific to orphan drugs and ensure patient input.</p>	<p>Department of Health.</p>
<p>There is a lack of information and guidance on the entitlements of patients who are refused funding for particular services or treatments/therapies.</p>	<p>6. Resources should be produced that inform patients of their rights and legal position if they are refused funding for treatments/therapies and how to go about the process of appealing a decision.</p>		<p>NHS Commissioning Board, (Health Watch England), Community Health Councils, Independent Advice and Support Services, Health Rights Information Scotland, Northern Ireland Patient and Client Council, patient organisations.</p>

## Patient Care, Information and Support

Current situation	Recommendations		Target of recommendation
<p>Patients are not provided with sufficient reliable information on their medical, psychological, social and other needs at diagnosis and throughout the progression of their condition.</p>	<p>1. Patients should be provided with ongoing, reliable information on their condition and how to manage it, which would include any existing treatment options, and how to receive the support they need.</p>		<p>a. "Information prescription" should be given on diagnosis.</p> <p>NHS, clinicians, commissioners, patient organisations.</p>
			<p>b. Reliable sources of information and patient organisations' contacts available to patients. (See also: Prevention and Diagnosis – Recommendation 3a).</p> <p>Health professionals.</p>
			<p>c. Post-diagnosis appointment with health professional to answer questions. (See also: Delivering Coordinated Care – Recommendation 3b).</p> <p>Clinical service managers, health professionals.</p>
			<p>d. Development of a referral facility to better assist patients in their search for information.</p> <p>Every NHS Trust/Health Board.</p>
			<p>e. Ensure patients access to information about treatment options in all stages.</p> <p>Healthcare professionals.</p>
			<p>f. Nominate a Care Coordinator responsible for answering to patients' questions. (See also: Delivering Coordinated Care – Recommendation 3b).</p> <p>Health departments, commissioners, clinical service managers, clinicians, patient organisations.</p>

Current situation	Recommendations	Target of recommendation
<p>Information to patients is not always provided in the appropriate format and at a level that ensures that the patient will be able to understand and use the content to support effective and appropriate decision making.</p>	<p>2. Information should be made available in various formats and at various levels of scientific and medical knowledge.</p>	<p>a. Fill in gaps in information available to patients.</p> <p>Health departments, patient organisations.</p>
		<p>b. Information should be available in a variety of formats.</p> <p>Health departments, patient organisations, community bodies including those representing BME communities and those with special needs.</p>
		<p>c. Links to reliable sources of information and guidelines in web portal. (See also: Prevention and Diagnosis – Recommendation 3a).</p> <p>UK health departments.</p>
		<p>d. Reduce bureaucratic barriers to good quality information.</p> <p>NHS Trusts/Boards.</p>
<p>Patients frequently report not being offered psychological support in relation to their condition.</p>	<p>3. Psychological support for the whole family should be considered an integral part of the care package.</p>	<p>a. Psychological and emotional support as part of patient’s care plan. (See also: Delivering Coordinated Care – Recommendation 2b).</p> <p>NHS, commissioners health professionals.</p>
		<p>b. Increase patient awareness of the various sources of support available. (See also: Recommendation 1a, 1b and 1d).</p> <p>Health professionals, NHS Trusts/Health Boards.</p>
		<p>c. To nominate a care coordinator as a point of contact. (See also: Delivering Coordinated Care – Recommendation 3b).</p> <p>Health departments, patient organisations.</p>
<p>Links between the medical and social aspects of care and support are often weak.</p>	<p>4. Social support for those affected by rare diseases should be a fundamental part of the patient’s care package.</p>	<p>See Recommendation 1a.</p>
		<p>See Recommendation 1b.</p>
		<p>See Recommendation 1d.</p>
		<p>See Recommendation 1f.</p>
		<p>See Prevention and Diagnosis - Recommendation 3a.</p>
		<p>See Delivering Coordinated Care -Recommendation 3a.</p>



# Delivering Coordinated Care

Current situation	Recommendations		Target of recommendation
<p>Care for patients with rare diseases is often poorly coordinated and fragmented, and there is frequently a lack of communication between all professionals involved in the care of the patient.</p>	<p>1. There should be a systematic programme of designation for centres of excellence for rare diseases supported by networks linking into local services throughout the UK.</p>	<p>a. To develop centres of expertise for groupings of rare conditions.</p>	<p>AGNSS, NSD, WHSSC, HSCB.</p>
		<p>b. Designated funding for specialised services. (See also: Commissioning and Planning – Recommendation 3a).</p>	<p>Health departments.</p>
		<p>c. Development of centres of excellence should be based on clinical needs. (See also: Commissioning and Planning – Recommendation 4b).</p>	<p>AGNSS, NSD, WHSSC, HSCB.</p>
		<p>d. Well-defined role of patient organisations.</p>	<p>AGNSS, NSD, WHSSC, HSCB, patient organisations.</p>
	<p>2. Mechanisms should be put in place to ensure good communication between all healthcare professionals involved in the care of a patient.</p>	<p>a. ‘Hub and spoke’ model between centres of excellence and local services. (See also: Prevention and Diagnosis – Recommendation 2c).</p>	<p>Commissioners.</p>
		<p>b. Personalised care plan for patients with chronic rare conditions.</p>	<p>Centres of excellence/ specialised services in collaboration with local services.</p>
		<p>c. Regular meetings between professionals involved in care of patients.</p>	<p>Clinical service managers, health professionals.</p>
		<p>d. ‘Flagging systems’ linking hospital admission systems and lead consultants.</p>	<p>NHS Trusts/Health Boards.</p>
		<p>e. Sharing hospital records.</p>	<p>NHS Connecting for Health.</p>
		<p>f. Patient-held medical records.</p>	<p>NHS Trusts/Health Boards, clinicians.</p>

Current situation	Recommendations		Target of recommendation
<p>Care for patients with rare diseases is not always patient-centred and does not fully take into account a patient's individual needs and preferences.</p>	<p>3. Care for patients with rare diseases should be patient-centred, taking account of an individual's personal needs.</p>	<p>a. To develop and support 'one stop shop' clinics.</p>	<p>Commissioners, centres of excellence, clinical service managers, health professionals, patient organisations.</p>
		<p>b. Patients should be offered a designated Care Coordinator.</p>	<p>Health departments, commissioners, clinical service managers, clinicians, patient organisations.</p>
		<p>See also Recommendation 2e and 2f.</p>	
<p>Care for patients with rare diseases is often not provided holistically and does not always include consideration of their non-medical needs.</p>	<p>4. Care for patients with rare diseases should be provided holistically, and should include consideration of the patient's and their family's non-medical needs.</p>	<p>a. Personalised care plan comprising psychological, social, financial support. (See also: Recommendation 2b).</p>	<p>Centres of excellence/ specialised services in collaboration with local services, care coordinators.</p>
		<p>b. Best practice guidelines to include transition from paediatric to adult services. (See also: Recommendation 1).</p>	<p>Centres of excellence/ specialised services, patient organisations.</p>
		<p>c. Needs of carers and families to be considered in the patient's care plan.</p>	<p>Centres of excellence/ specialised services in collaboration with local services, care coordinators.</p>
		<p>See also Recommendation 3b.</p>	
		<p>See also Recommendation 2f.</p>	
		<p>See also Patient Care, Information and Support – Recommendation 1a.</p>	

# **Improving Lives, Optimising Resources:** A Vision for the UK Rare Disease Strategy

Findings in depth

# Coordination of Research

## Current situation

1. There is limited collaboration between researchers working on rare diseases.
2. There is limited funding provision for research into rare diseases.
3. There is a lack of basic epidemiological and clinical data on rare diseases.
4. There are difficulties in the development of new diagnostic tests for rare diseases.
5. The need to obtain Research and Development (R&D) approval from numerous sites slows and inhibits rare disease research.
6. There are difficulties in therapeutic and prevention research, including research methodologies applicable to rare disorders and the development of orphan drugs for children.
7. There is currently very little research on the most effective management of patients with rare diseases and management guidelines exist for very few disorders.

## Recommendations

1. Mechanisms should be put in place to improve collaboration between rare disease researchers.
2. Funding bodies should be encouraged to support research into rare diseases by provision of specific funding streams.
3. Support should be given to develop and sustain systems for data collection and disease registries for patients with rare diseases.
4. Measures should be taken to encourage the development and approval of diagnostic tests.
5. The system for gaining R&D approval for research that spans the UK should be streamlined.
6. Major funding bodies should be encouraged to explore and promote research and debate into appropriate and acceptable research methods into the prevention and treatment of rare disorders.
7. Funding bodies, including the NIHR, should commission research on health service delivery for patients with rare disorders, and promote and support the development of guidelines as tools to improve care management.

## Limited collaboration and funding in rare disease research

### Current situation

**There is limited collaboration between researchers working on rare diseases.**  
**There is limited funding provision for research into rare diseases.**

The research base for individual rare diseases is often limited to a small number of individuals spread both nationally and internationally. Without formal networks in place to connect researchers, collaboration may be limited resulting in duplication of effort, inappropriate competition for funding and, overall, a lack of strategic direction.

The UK currently does not participate in E-Rare, a network of 16 partners from 12 European countries responsible for the development and funding of national and regional rare disease research projects. E-Rare partners exchange information on rare diseases and organise joint funding for research projects. The lack

of contribution from the UK means that there are fewer opportunities for UK researchers to collaborate, leading to reduced success in funding proposals.

Overall, the amount of funding provided for research into rare conditions in the UK is limited. Although it is not possible to obtain exact figures of the total funding for rare disease research, the Association of Medical Research Charities (AMRC) estimates that in 2008/09 charities invested approximately £3.6 million in rare disease research, compared to £370 million in cancer research. Based on best estimate figures suggesting that 3.5 million people are affected by rare diseases, this equates to £1 of charitable funding per patient, compared to £185 per patient for cancer (based on 2 million affected people)<sup>5</sup>. On top of this, there is often a disproportionate reliance on charitable funding for rare disease research, indicating that major national funders do not include research into rare disease as a priority and are often reluctant to support such research because of a perceived lack of impact on the burden of disease and expected limited cost-effectiveness due to the small number of affected people. Large research funding bodies do not allocate the proportion of funding to rare diseases that the number of affected people might justify. Absence of dedicated funding streams for rare diseases also results in inequity of research between conditions, often dependent on the activity of patient organisations to raise funds for such research.

Without a systematic programme of research, progress in the development of diagnostic tests and treatments for rare conditions is greatly hindered.

### **Recommendation 1**

#### **Mechanisms should be put in place to improve collaboration between rare disease researchers.**

Enabling and encouraging researchers to be aware of each other's work and to work together more effectively would result in the limited funds for research into rare conditions being deployed more systematically and strategically, and would also help to draw in available collaborative expertise.

#### **Specific actions that should be taken to improve collaboration between researchers include:**

- a. The development of, and strategic funding for, clinical research networks (CRNs) focused on rare disorders. A CRN is a multidisciplinary multi-centre network, with agreed aims and objectives, researching a particular condition or group of conditions. Support for the development of these networks would enable experts to collaborate nationally and internationally, leading to a pooling of knowledge and expertise. Clinical research networks can inform best practice, develop care guidelines and protocols, and ultimately lead to an improvement in the lives of patients. Research by such networks would also result in savings to the NHS, due to improved diagnosis and more effective and appropriate treatments and management of patients. Although there may need to be some initial investment in setting up CRNs, we believe that over time the benefits and savings from these initiatives would outweigh this input.

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5. '2009 Annual Report of the Chief Medical Officer', Department of Health, March 2010

## Benefits of an effective clinical research network: an example of good practice

The Treat-NMD network for neuromuscular diseases (<http://www.treat-nmd.eu/home.php>) is an example of an effective collaborative network. It was established in 2007 thanks to a five-year EU funding grant, and aims to advance diagnosis, care and the development of new treatments for those affected by all inherited neuromuscular diseases. The network now has 22 interested partners covering 11 countries with more than 300 collaborators worldwide. The services and tools provided by this network include:

- the production and dissemination of accredited care standards and family guides on Duchenne muscular dystrophy and spinal muscular atrophy
- a registry of outcome measures
- a communications infrastructure
- a network of care and trial sites
- registries of patients with neuromuscular diseases
- training and education of professionals
- a DNA, cell and tissue biobank.

This work has a direct beneficial impact on patients affected by these conditions and would not have been achieved without the collaboration made possible due to this network. We recommend that Treat-NMD be used as a model of good practice when developing similar networks.

- b. Established research networks, such as the NIHR Comprehensive Clinical Research Networks, should recognise that they have a responsibility for rare diseases and should support research into appropriate rare disorders as part of their work plan. If working effectively, networks lend themselves to rare disease research.
- c. The expertise of existing networks should be utilised to help in the development of new, smaller networks. This might be, for example, by utilising expertise from sources such as the NIHR Clinical Genetics Network in database development or advice on structure and management of the network. The NIHR Comprehensive Clinical Research Networks work to provide a focus for multiple topics of research, resulting in well-developed models of communication and interaction between researchers. We recommend that the Clinical Genetics Network expand its remit to include all rare diseases, so that researchers in this field are also able to benefit from its knowledge and successful systems.

## Recommendation 2

### Funding bodies should be encouraged to support research into rare diseases by provision of specific funding streams.

Apart from its prime impact on the understanding and management of rare disorders themselves, research into rare conditions may also result in insights into the pathogenesis of more common conditions by identifying developmental pathways or mechanisms of disease causation. However, this is often not appreciated by large research funding bodies, which focus their support on large scale studies of complex diseases.

#### Specific actions that should be taken to ensure rare diseases are given more consideration by funders include:

- a. Continual promotion of the importance of rare disease research to large funding bodies such as the MRC and NIHR. We recommend the inclusion of someone with rare disease expertise on various committees of the funding boards, in order to raise awareness of the issue and ensure that adequate thought and consideration is given to applications from rare disease researchers.
- b. Awareness needs to be raised of the high quality research that is currently being supported in the



UK by smaller charitable funding. Collaborations between patient organisations and researchers are often limited by resources, however, and could be supported by grants from larger funders. Increasing funders' knowledge of the important research that is taking place into rare diseases would make them aware of the potential high returns from their input and encourage them to get involved. Organisations such as the UK branch of Orphanet could help in raising awareness of the research that is being conducted into rare diseases.

- c. The UK should participate in E-Rare. For many rare conditions, international collaboration is required for effective research to be carried out, and not participating in this programme hinders the UK's ability to make progress in rare disease research.

## Lack of effective methods of data collection

### Current situation

#### **There is a lack of basic epidemiological and clinical data on rare diseases.**

Basic epidemiological data and information on the natural history of many rare diseases is very limited. However, this knowledge is essential in making the case for funding and provides a basis for many research studies, particularly those assessing the possible impact of preventive or treatment activities. Effective systems for collection of data would also help to identify patients who may wish to participate in clinical trials. Patients are generally very willing to be involved in research, so there must be better systems in place to ensure that they are informed of ways in which they can participate.

In October 2010 the Department of Health (DH) published a consultation document on 'An Information Revolution,'<sup>6</sup> that states that the government wants to transform 'the way information is collected, analysed and used by the NHS' in England and discusses the need to capture data accurately. We agree this is important particularly for patients with rare diseases and initiatives within all four home nations should be brought together to allow a UK-wide approach to data collection and use.

### Recommendation 3

#### **Support should be given to develop and sustain systems for data collection and disease registries for patients with rare diseases**

##### **Specific recommendations to improve rare disease data collection include:**

- a. The development of disease registries; these should be promoted by the national health systems in all four home nations, patient organisations, funding bodies, industry, researchers and clinicians through clinical and research networks. A disease registry is a database that collects clinical information from all patients with a particular condition, or type of condition. Registries can provide essential information for clinical care, planning and service delivery, and are a valuable tool for the initial collection of data on rare disease patients. Healthcare professionals should therefore provide thorough information about registries to their patients and encourage them to join. With consent from patients, researchers can also use registries to identify and recruit appropriate patients into clinical trials, to study epidemiology and natural history, and to evaluate and audit patterns of service delivery.

6. [http://www.dh.gov.uk/en/Consultations/Liveconsultations/DH\\_120080](http://www.dh.gov.uk/en/Consultations/Liveconsultations/DH_120080)

### Benefits of a registry: an example of good practice

The European Huntington's Disease (HD) registry (<http://www.euro-hd.net/html/registry>) is an example of a successful registry. It was established as part of the European Huntington's Disease Network (EHDN) in 2003.

So far 19 countries and 140 study sites participate in this registry, which currently contains details of almost 7,000 HD patients and family members. The aims of this registry are:

- To collect natural history data of patients.
- To relate genetic mutation with clinical symptoms.
- To identify and recruit appropriate patients for clinical trials.
- To plan future research studies.
- To develop new ways to track and predict disease onset and progression.

This registry contributes to the development of deeper understanding of HD, including improving knowledge of factors that have an impact on the onset, symptoms and cause of HD. It also keeps patients informed of relevant trials and research in which they can participate.

This registry is guided by a steering committee consisting of one clinician from each participating country and key representatives from the EHDN. This has proven an effective way of managing a registry.

This registry could be used as a model on which to base further rare disease registries.

- b. We recommend that funding bodies should regard support for infrastructure of registries as being equal in importance to the actual research.

It is essential in the setting up of disease registries that due consideration is given to ensuring long-term sustainability as it may take time to acquire sufficient data for the registry to be of optimal use. Consideration must also be given to the ownership and management of the registry and access arrangements to the registry content.

- c. There needs to be increased collaboration and harmonisation of data sets to allow pooling of more data (for example from other regions or countries) in order to identify larger cohorts of patients for research and enable further research into the underlying features of the disease. Any new data collection initiatives should be harmonised with existing registries as far as possible.

We therefore recommend that a symposium of all funders of such work be developed to discuss commonality and future sharing and interoperability of data sets. This should be included by government as part of their Information Revolution work, and should combine initiatives across the UK.

- d. Sustainable funding for surveillance units should be made available to ensure that they can continue to work as effectively as possible.

Rare disease surveillance units are another effective way of collecting epidemiological data. Data from surveillance units can assist research, as well as influencing public health policies. The UK is fortunate in having well-defined rare disease surveillance networks, such as the British Paediatric Surveillance Unit (BPSU). The BPSU has been in existence for 25 years and has been funded by the DH for the past 12 years. Its methodology, adopted internationally, allows for multi-national data collection and comparison. However the BPSU is not comprehensive and relies on the research initiatives of participants. There are now other surveillance units in a variety of specialties operating in all four home nations. These units, although they work separately, are able to combine when necessary to maximise case ascertainment. The information gathered allows for the assessment in real time of disease presentation and treatment, as well as looking for regional clusters and cultural differences. However, like registries, these units have difficulty in obtaining long-term secure funding.

- e. Timely implementation of the International Classification of Diseases (ICD) 11, which offers greater granularity in rare disease classification, and will provide opportunities for capturing data on the

incidence and natural history of rare diseases.

The UK's National Health Service is almost unique worldwide in being able to support clinical research as it is a healthcare system for more than 60 million people. UK-wide, central collection of the right type of data can facilitate research and development, and is essential for patients with rare diseases. However, the current coding system is ineffective for rare diseases, as the breakdown of conditions is too broad and important details cannot be captured.

- f. The development and ongoing support of databases that collect data on disease genotype/phenotype correlations. DECIPHER<sup>7</sup> is an example that receives contributions by scientists and clinicians worldwide in relation to chromosomal imbalances related to developmental delay. It has enabled the delineation of new conditions and their underlying genetic mechanisms. Access to this database by clinicians also greatly aids clinical management since individual data would not have been published and could not be obtained through any other route. Again, sustainability of funding needs to be ensured.

Recommendation 1 would also aid the collection of epidemiological data on rare diseases.

## Development and implementation of diagnostic tests

### Current situation

#### **There are difficulties in the development of new diagnostic tests for rare diseases.**

Historically, difficulties have been experienced mostly because setting up new diagnostic tests using current technology can be time-consuming and costly, and, because of the small target population the tests may not be seen as cost-effective. However, rapid progress in genetic technology, notably the innovative application of Next Generation Sequencing (NGS) platforms will impact significantly on the speed with which new and rare genetic tests can be developed, although their validation will need careful implementation.

The capacity and resolution of continually improving NGS platforms will also mean that clinical scientist skill mixes will need to include comprehensive bioinformatics training so that NGS technology can be fully exploited. It is currently uncertain exactly how NGS technology will be deployed within the health service but it is becoming clear that public/private partnerships will emerge as potentially significant players and that some rationalisation of the number of Regional Genetics Laboratories providing genetic testing for rare disorders may be necessary.

Further modernisation and rationalisation of laboratory genetics services are being driven by (a) the implementation of an integrated genetics and pathology clinical scientist training programme under the DH's Modernising Scientist Careers initiative, and (b) the Association of Clinical Cytogenetics and Clinical Molecular Genetics Society, which are moving towards forming a single professional body, thereby effectively ending the distinction between the current "cytogenetics" and "molecular cytogenetics" disciplines. Taken together, it is to be hoped that these initiatives will facilitate the development and implementation of genomic technologies and further enhance an already strong collaborative ethos within all the genetics disciplines including clinical genetics. It will also continue to be important to maintain significant clinical genetics input into the application and appropriate use of new tests as they become available.

The increasing diagnostic application of technologies such as array comparative genomic hybridisation (array-CGH) and the forthcoming UK-wide collaborative study coordinated by the Sanger Centre in Cambridge - Deciphering Developmental Disorders (DDD) - will undoubtedly result in a significant increase in the detection of pathogenic genomic (by array-CGH) and molecular (by exome resequencing) mutations in patients with currently unexplained neurodevelopmental disorders. The DDD may also provide a unique opportunity for a research forum focused on groups of patients with rare diseases.

Current commissioning and planning arrangements can be slow to respond to requests to fund an expanding repertoire of genetic tests. At present this involves a process of evaluation of the analytical and clinical validity and clinical utility of genetic tests by the UK Genetic Testing Network (UKGTN). This is a very robust and evidence-based system for tests for rare inherited conditions where patients are referred to clinical genetics. Tests that pass evaluation are recommended to commissioners. This process does not evaluate

7. <https://decipher.sanger.ac.uk/application>

the technology but will consider whether the methodology proposed is appropriate e.g. utility of arrayCGH for diagnosis of causes of learning disability or developmental delay with dysmorphism or congenital malformations. UKGTN also provides an extended advisory role for tests that do not meet their remit e.g. UKGTN recently provided advice to commissioners on non-invasive prenatal diagnosis for sex linked conditions. The Westminster Government's recent White Paper<sup>8</sup> proposes fundamental changes to NHS funding in England but the impact of these changes on specialist commissioning arrangements is not yet clear.

Development of non-genetic tests, including biomarker analysis and imaging assessments, can also be difficult. The NIHR currently excludes direct funding for laboratory-based projects for the development, testing and evaluation of novel genetic tests. However, it has been developing other funding streams including the NIHR Health Technology Assessment (HTA) and the MRC's Efficacy and Mechanism Evaluation (EME) programmes, which could be applied to the development of new tests with defined research protocols. The Chief Scientific Officer for England also runs annual Healthcare Scientists Research Fellowship competitions, one of the stated goals of which is 'the development of new and improved diagnostic testing'

## Recommendation 4

### Measures should be taken to encourage the development and approval of diagnostic tests.

- a. There needs to be a coordinated national strategy for the development of rare genetic tests in order to avoid duplication and to develop consortium approaches. An example of effective working is the SCOBEC Genetics Laboratory Network, a collaboration of six molecular genetics laboratories in the UK which have been successful in increasing the capacity and reducing the costs of genetic tests. Rationalisation and coordination of laboratory services will be particularly important in the context of the use of NGS.
- b. The UKGTN gene dossier process has an excellent record for the evaluation and recommendation of genetic tests for rare inherited conditions. The process by which UKGTN approval actually translates into laboratory funding however does not work well and serious consideration needs to be made as to how this could be improved. Nevertheless, Regional Genetics Laboratories should fully engage with the UKGTN to expand the repertoire of approved dossiers and increase the number of available testing criteria which promotes appropriate ordering of genetic tests.
- c. There are a number of NIHR funding streams for the evaluation of laboratory aspects of the development of genetic tests (NIHR, HTA and EME). Most of these require coordinated research projects which in turn need the further engagement of academic, clinical and laboratory genetic professionals to exploit these opportunities. The development of high throughput technologies for rare diseases poses particular problems and there is a need for coordinated approaches that include specific funding for laboratory developmental work.
- d. The forthcoming DDD project may offer a unique opportunity for the use of ultra high resolution array-CGH and exome resequencing on defined groups of consented patients with rare diseases.

## Difficulties in obtaining approval for research

### Current situation

#### The need to obtain Research and Development (R&D) approval from numerous sites slows and inhibits rare disease research.

Research into rare conditions often requires recruitment of patients from multiple different sites, due to the often low numbers of people affected by a particular rare condition. The Integrated Research Application System (IRAS) has streamlined and speeded multi-site Research Ethics Committee (REC) approval. This streamlining has also improved the efficiency in gaining National Information Governance Board (NIGB) approval when identifier data is collected without consent. However, there is still a barrier in gaining local Research and Development (R&D) approval because under the current system approval must be gained from the site where the research is to be undertaken and each individual local R&D office needs to be informed. This is compounded by the fact that there is no standard form between sites and the requirements vary for each, and that no standard R&D office contact listing exists. The result is often a very time-consuming and

8. 'Equity and excellence: Liberating the NHS'; Department of Health; July 2010.  
[www.dh.gov.uk/prod\\_consum\\_dh/groups/dh\\_digitalassets/@dh/@en/@ps/documents/digitalasset/dh\\_117794.pdf](http://www.dh.gov.uk/prod_consum_dh/groups/dh_digitalassets/@dh/@en/@ps/documents/digitalasset/dh_117794.pdf)

challenging process that can hinder the progression of research into rare diseases either by delaying the onset of research projects, or causing termination of a project as a result of a lack of available resources to go through the approval process. The burden of obtaining all necessary approvals may also result in research being driven abroad where the equivalent requirements are often simpler to obtain. Rare diseases are particularly affected by the current regulatory requirements, as projects are often small and have limited funds to allocate to obtaining the appropriate approvals, as well as the need for patients to be recruited from multiple sites. This could be mitigated in part if rare disease research could be considered as falling under the NIHR Comprehensive Clinical Research Networks in England, the National Institute for Social Care and Health Research Clinical Research Centre in Wales, the Scottish Academic Health Sciences Collaboration and the relevant body in Northern Ireland.

RDUK has been made aware of numerous examples of the difficulties of obtaining all the required approval to begin research projects. Two examples of difficulties are:

- a DH-funded project supported by all four UK Chief Medical Officers that sought fast track approval in view of the public health urgency of the study. The process was started in July 2009, the ethics and NIGB approval were speedily given, but due to the complexities of R&D approval, and despite having two full-time experienced researchers working on this from the start, the required UK R&D consents were not all received until February 2010. In contrast, a similar project in Australia was able to get consent and begin the study within 10 days.
- The RAPID (Reliable Accurate Prenatal non-Invasive Diagnosis) project<sup>9</sup> had to wait over a year before they had R&D approvals for all their sites.

NIGB approval for the unconsented collection of identifier data does not go beyond England and Wales. In Scotland and Northern Ireland there is currently no statutory requirement to get such additional consent, however funders may require such approvals and further delays and extra costs may occur when seeking approval from individual NHS organisation Caldicott Guardians.

The fees charged by R&D departments are not standardised and vary considerably. In many countries in Europe, R&D fees are not applied to independent research projects by Academic Institutions. However, in the UK, independent studies can have R&D fees charged for conducting the research in that Institution and these have been known to be as high as 45% of the total study budget. This considerably increases the funding needed to conduct independent research, which is of concern in rare disease research where funding is already limited.

## Recommendation 5

**The system for gaining R&D approval for research that spans the UK should be streamlined.**

**Specific actions that should be taken include:**

- a. A reappraisal of the local R&D approval system should be undertaken to consider blanket approval, or at least the standardisation of approval forms to minimise the time spent seeking approval.

Following the DH White Paper 'Equity and excellence: Liberating the NHS'<sup>10</sup>, the Academy of Medical Sciences was commissioned to carry out an independent review of the regulation and governance of medical research. The DH stated that 'in light of this review we will consider the legislation affecting medical research, and the bureaucracy that flows from it, and bring forward plans for radical simplification'.

The Academy of Medical Sciences published its review 'A new pathway for the regulation and governance of health research'<sup>11</sup> in January 2011. It included recommendations such as that there needs to be 'rationalisation of the regulation and governance of all health research' including a single system for ethics review. It also highlighted the importance of 'providing access to patient data that protects

9. [www.rapid.nhs.uk](http://www.rapid.nhs.uk)

10. [www.dh.gov.uk/prod\\_consum\\_dh/groups/dh\\_digitalassets/@dh/@en/@ps/documents/digitalasset/dh\\_117794.pdf](http://www.dh.gov.uk/prod_consum_dh/groups/dh_digitalassets/@dh/@en/@ps/documents/digitalasset/dh_117794.pdf)

11. 'A new pathway for the regulation and governance of health research', The Academy of Medical Sciences, January 2011 [www.acmedsci.ac.uk/index.php?pid=47&prid=88](http://www.acmedsci.ac.uk/index.php?pid=47&prid=88)



individual interests and allows approved research to proceed effectively' and of 'embedding a culture that values research within the UK'.

We support this review, in particular the recommendation that 'changes are needed to reduce bureaucracy and increase the speed of NHS R&D permissions by replacing multiple, inconsistent, slow checks by individual NHS Trusts, with a single, consistent, efficient process for the NHS as a whole'. We recognise that the government is therefore now aware of the issues surrounding approval for research, and we feel it is important that following the review they consider the impact on rare disease research when developing their plans in this area.

- b. There is the need for R&D approval to cover all four home nations. Research Ethics Committee (REC) approval is streamlined and centralised for the whole of the UK and NIGB approval is now centralised for England and Wales. Both of these systems make getting approval much simpler than for R&D. If it has been possible to put processes in place to make this work for other approval systems, we believe it should be possible to do the same in terms of R&D to further improve research into rare diseases.
- c. Requirements for R&D approval for research into rare diseases need to be much more in line and proportionate to the research project being carried out. Although we appreciate the reasoning behind the need for approvals, for some small or low cost research projects the lengths that have to be gone to are not in line with the size and complexity of the project. Processes need to be put in place that make it simpler to get approval for projects that do not involve invasive procedures or trials on patients or those without major ethical issues. Groups such as the Human Genomic Strategy Group and the NIHR Clinical Genetics Network should be asked to investigate this further.
- d. R&D fees should be standardised between sites in the UK and must be made more transparent to help facilitate rare disease research. The fees should also be more proportionate to the size and complexity of research projects, and should be in line with the requirements for R&D approval.

## Research into prevention and treatment

### Current Situation

**There are difficulties in therapeutic and prevention research, including research methodologies applicable to rare disorders and the development of orphan drugs for children.**

There are currently difficulties in initiating research to develop and evaluate effective diagnostic, preventive and treatment options in rare disorders. The emphasis placed by funding bodies and those responsible for health technology assessment on the importance of evidence from randomised control trials (RCTs) presents a significant obstacle for rare disorders where it may be difficult or impossible to gather sufficient numbers of patients and generate the necessary levels of statistical significance. Paradoxically, the very level of detail that it is possible to know about disease in patients suffering from rare disorders may generate an even more stringent set of standards when evaluating effects of preventive measures or treatment, for example by requiring that outcomes are compared for the various underlying subsets (e.g. particular mutation in genetic disorders, or enzyme level in inherited metabolic disease).

A final problem with respect to drug treatments is that pharmaceutical companies are often reluctant to trial drugs on children until sufficient research is done on adults. However, due to the severity of the conditions many children affected by rare diseases die before they reach adulthood, meaning that sufficient research on adults can rarely be achieved.

In 2007 the Paediatric Regulation<sup>12</sup> was introduced by the European Medicines Agency (EMA). The aim of the Regulation was to ensure that the development of medicines for children was included as part of the mainstream drug development process. The Regulation requires pharmaceutical companies to produce a Paediatric Investigation Plan (PIP) to be submitted to the Paediatric Committee (PDCO) as part of the marketing authorisation process for a new drug. The PIP should detail how the company proposes to investigate the drug for use in children. In terms of drugs for rare diseases, the regulation states that 'the ten-year period of orphan market exclusivity should be extended to twelve years if the requirement for data on use in the paediatric population is fully met'. As the majority of rare diseases affect children, it is very important that the regulation for the development of a PIP is adhered to in the development of orphan drugs.

12. [http://ec.europa.eu/health/files/eudralex/vol-1/reg\\_2006\\_1901/reg\\_2006\\_1901\\_en.pdf](http://ec.europa.eu/health/files/eudralex/vol-1/reg_2006_1901/reg_2006_1901_en.pdf)



## Recommendation 6

**Major funding bodies should be encouraged to explore and promote research and debate into appropriate and acceptable research methods into the prevention and treatment of rare disorders.**

This is a necessary step because conventional research methods may not be appropriate for rare disorders due to small populations. The research and debate should take into account the overall restriction in patient numbers and the heterogeneity within patient cohorts. It should also recognise the different ways in which patients with rare disorders can be studied, and, in particular, the opportunities for detailed observational studies of pathological processes and how they can be influenced at a molecular level.

## Patient management research

### Current situation

**There is currently very little research on the most effective management of patients with rare diseases and management guidelines exist for very few disorders.**

This situation is partly due to the rarity of the disorders and the fact that even specialist centres may have relatively few affected patients. The work itself is time-consuming and requires widespread professional ownership, often on an international basis. It has therefore largely taken place in the context of clinical areas where there are active professional clinical networks. Development of evidence-based guidelines may be difficult because of the lack of evidence but methodologies do exist for maximising the evidence that is available.

The guidelines should not only cover details of individual patient assessment and treatment, but also recommended organisational aspects of management such as referral protocols and local delivery of care under specialist supervision. Such research has the potential to streamline systems, save NHS money and improve patient outcomes.

### Recommendation 7

**Funding bodies, including the NIHR, should commission research on health service delivery for patients with rare disorders, and promote and support the development of guidelines as tools to improve care management.**

# Prevention and Diagnosis

## Current situation

1. There is a lack of awareness and identification of rare diseases among healthcare professionals, often resulting in a delay in diagnosis or misdiagnosis of rare disease patients.
2. It can be difficult both for professionals and patients to access reliable, up-to-date information on rare diseases and who the specialist(s) is/are in a particular condition.
3. The criteria currently used by the National Screening Committee (NSC) to determine suitability for newborn screening tend to militate against rare conditions.
4. There is inequity of access to diagnostic tests across the UK.

## Recommendations

1. Increase healthcare professionals' knowledge and awareness of rare diseases.
2. There is a need for improved linkage between specialist centres and local services to enable education of local healthcare professionals.
3. Improve access to reliable information on rare diseases to make it easier for the public and professionals to obtain information.
4. Appropriate rare diseases need to be considered for inclusion in the newborn screening programme.
5. There must be improved access to diagnostic tests in under-served areas of the UK to ensure equity of access throughout the country.
6. Access to carrier tests for individuals and groups considered to be at significant risk of a specific condition should be facilitated and promoted.

## Lack of awareness of, and information on, rare diseases

### Current Situation

**There is a lack of awareness and identification of rare diseases among healthcare professionals, often resulting in a delay in diagnosis or misdiagnosis of rare disease patients.**

The consequence is a delay in accessing appropriate treatment, therapy or effective management of the condition, as well as an inefficient use of NHS resources due to multiple avoidable appointments with different consultants and incorrect diagnostic tests and treatments. A delay in diagnosis can cause a reduction in the patient's life expectancy and quality of life, while a misdiagnosis may result in a patient being managed for a condition - often a more common condition - that they do not have. Delays in diagnosis can result in missed opportunities for intervention, allowing conditions to become progressively worse and more difficult - sometimes impossible - to treat. For example, the response of a patient to enzyme replacement therapy is optimised the earlier the patient starts therapy. Early diagnosis, even for untreatable conditions, can also provide important information to guide future reproductive choices for the family.

RDUK has encountered numerous examples where there have been issues in obtaining a diagnosis of a rare condition. The Behçet's Syndrome Society, for example, has found that the average wait for a diagnosis of Behçet's syndrome is 12 years from the onset of symptoms. The Society is also aware of cases where this wait

has resulted in the death of patients due to delayed access to appropriate medication<sup>13</sup>. The British Thoracic Society has said that problems in diagnosis are 'too numerous to mention'<sup>14</sup>, and the ChILD Lung Foundation also reports that it commonly encounters problems in diagnosis<sup>15</sup>.

Research conducted by RDUK for its recent report on patients' and families' experiences found that 46% of rare disease patients had to wait over a year for a final diagnosis following the onset of disease symptoms. Of these patients, 20% had to wait over five years, and 12% had to wait over ten years. RDUK's research also showed that 46% of patients were given an incorrect diagnosis, of which 30% received three or more incorrect diagnoses prior to receiving a final diagnosis of a rare condition.

Measures are needed to improve awareness of rare diseases among healthcare professionals to increase the likelihood of a rare condition being considered among the differential diagnoses at an early consultation. As a result, earlier diagnosis and prompt access to necessary services and treatments would be facilitated.

Clinicians reasonably suspect that unusual symptoms in patients are often the result of an unusual presentation of a common condition. With greater awareness and access to better information, clinicians are more likely to be able to identify when this is not the case and a patient in fact has a rare condition.

## Recommendation 1

### **Increase healthcare professionals' knowledge and awareness of rare diseases.**

It is clearly not possible for health professionals to know about all rare disorders. However, clinicians must develop strategies that make earlier consideration of referring patients to specialist consultants more likely, when referral is appropriate and there is a significant probability that the patient has a rare condition.

#### **Specific actions that need to be taken to increase awareness and knowledge of rare diseases among healthcare professionals include:**

- a. Many rare disorders have a genetic origin and accompanying multisystem effects. There are therefore some simple guidelines such as a family history or multiple apparently unconnected signs which may trigger the consideration of a rare condition. This type of basic teaching must be included in the curriculum for medical students and other healthcare professionals in training. A module that identifies pointers to the possibility that a patient has an uncommon condition would alert trainees to the existence of rare conditions and introduce the steps in their proper investigation. This module should cover general principles involved in diagnosing a patient with a rare disease, such as the appropriate aspects of the clinical and family history to question, the initial investigations to undertake, when to refer and to whom, together with sources of information on rare diseases or tests for rare diseases. It should not provide information on all rare conditions, but instead be a more generic approach with some case studies of patients' experiences with rare diseases to provide evidence of their importance.

Training of this type would help to improve the diagnosis of rare conditions and would make a great difference to the lives of patients with these conditions. As such, it should be a mandatory part of training, particularly for paediatricians. We believe that a module of this sort would be attractive to practitioners as many are aware that it is an area in which they lack knowledge and would appreciate well-structured training.

- b. Ongoing training on rare conditions should be provided through well-designed modules as part of CPD/ CME (Continuing Professional Development/Continuing Medical Education) for healthcare professionals. This training would enable professionals to build on the foundations that they are taught as trainees, or to learn new skills if they have not previously been taught about rare conditions. Professionals could opt in to additional training on topics that they personally felt they needed to learn more about, and the CPD points would act as an incentive for them to dedicate time to this.
- c. As the Department of Health (DH) White Paper 'Our Inheritance, Our Future'<sup>16</sup> indicated, the training of secondary healthcare professionals (those medical specialists to whom patients are referred by a primary care provider such as a GP) in basic genetics should be improved. As 80% of rare conditions

13. Bechets Syndrome Society response to RDUK consultation, Oct 2010

14. British Thoracic Society response to RDUK consultation, Oct 2010

15. ChILD Lung Foundation response to RDUK consultation, Oct 2010

16. 'Our Inheritance, Our Future. Realising the potential of genetics in the NHS', Department of Health, June 2003

have a genetic origin, ensuring an underlying, fundamental understanding of genetics among secondary healthcare professionals would raise awareness of a considerable number of rare conditions and their inheritance patterns. However, it is equally important that education on non-genetic rare diseases is also provided.

The National Genetics Education and Development Centre<sup>17</sup> is a DH and Welsh Assembly Government-funded centre that works to facilitate the integration of genetics into the education of healthcare professionals. The Centre runs events to educate healthcare professionals on genetics and the impact that genetics can have on health. It also provides online links to other useful resources such as 'Telling Stories', which was developed to promote understanding of genetic conditions for the nursing profession based on real-life examples. Although not all rare conditions are genetic, projects such as this are useful and reliable sources of information that can help to raise awareness of these conditions to the relevant healthcare professionals. Similar initiatives by health departments to cover a wider range of rare diseases would be very helpful to develop this successful approach.

- d. The development of e-learning packages on rare diseases would assist in the training of qualified medical professionals. These should be developed by the National Genetics Education and Development Centre, in partnership with the Royal Colleges under the guidance of the Joint Committee on Medical Genetics, and should aim to increase overall awareness of rare diseases, rather than focussing on recognition of particular specific disease symptoms.
- e. Currently it often falls to teams from the Regional Genetics Services to educate other healthcare professionals on rare diseases. Where this is the case, it is important that this educational role is recognised and allocated as a formal part of their job planning.

### Production of educational material: an example of good practice

Increased awareness and knowledge will ensure that rare diseases are considered when a diagnosis is being made, therefore facilitating the diagnostic process. As an example, the UK Primary Immunodeficiency Network<sup>1</sup> (UKPIN) has produced educational material for primary and secondary care physicians. UKPIN has developed a set of national guidelines on the diagnosis and referral of primary antibody deficiency<sup>2</sup> and has led the development of a series of diagnostic reminders aimed at various specialists including respiratory, ENT (ear, nose and throat) and paediatrics<sup>3</sup>. It will be carrying out an audit of the impact of these educational campaigns. This example of good practice is an initiative that could be extended to other conditions.

<sup>1</sup> [www.ukpin.org.uk](http://www.ukpin.org.uk)

<sup>2</sup> Primary antibody deficiencies: recognition, clinical diagnosis and referral of patients. Wood P, UKPIN, Clin, Med., December 2009, 9(6): 595-9

<sup>3</sup> [www.pia.org.uk/includes/documents/DiagnosisGuides.pdf](http://www.pia.org.uk/includes/documents/DiagnosisGuides.pdf)

## Recommendation 2

### There is a need for improved linkage between specialist centres and local services to enable education of local healthcare professionals.

Currently the diagnosis of a rare condition can depend to a large extent on geographic proximity to the specialist centre for that condition. It is often the case that the closer an individual lives to a specialist centre, the more likely they are to come to attention and be diagnosed if they have that condition. Clinicians in local services must be able to recognise the limitations of their own knowledge and when they need actively to seek help and guidance from specialist centres and experts. The process of seeking specialist support should be streamlined to make it as simple as possible to identify the appropriate expert(s) and to liaise with them.

#### Specific actions that need to be taken to improve linkage include:

- a. Staff exchanges, whereby staff from specialist centres undertake outreach work and educate staff at local services to share up-to-date and relevant information on best practice care for the rare conditions in which they specialise. Such an initiative takes place at the Royal Free Hospital in London, where scleroderma outreach clinics carried out by the rheumatologist and the cardiac consultant ensure that the

<sup>17</sup> [www.geneticseducation.nhs.uk](http://www.geneticseducation.nhs.uk)

care of patients is shared between the specialist centre and the regional district general hospitals.

Immunology centres also work closely with local hospitals and GPs to provide good quality, local-based care for patients with primary immunodeficiency diseases.

The role of patient organisations in this type of education must also be noted. Where there is no specialist clinic for a condition, it is often the patient organisation that has the most knowledge. Funding should be available for these organisations to educate healthcare professionals at local hospitals, integrated with NHS-led CPD/CME.

### The role of patient organisations in education: an example of good practice

The Jennifer Trust for Spinal Muscular Atrophy has an Outreach Service (funded by the Big Lottery Fund) which allows them to employ two outreach workers. Their role is to provide a key service to newly diagnosed and bereaved families, but also to arrange local training sessions for health professionals so that the families can be better supported in the local community.

- b. Staff from specialised services should have an educational role included as part of their job description, and as such should dedicate an agreed amount of time to educating local staff. This could include input to courses and other teaching activities, as well as staff exchanges. Again, where there is no specialist centre, patient organisations may perform a valuable role and there should be mechanisms in place to ensure sustainable funding for them to do so.
- c. A 'hub and spoke' model should be developed in which specialist centres feed information to local clinics. Effective commissioning and planning structures could create the framework to achieve this necessary integration. It should be mandatory that the Advisory Group for National Specialised Services (AGNSS) in England, the Welsh Health Specialised Services Committee in Wales, the National Services Division in Scotland, and the Health and Social Care Board in Northern Ireland give attention to linkage between specialised and local services, and actively plan how to achieve this when commissioning and developing specialised services.

### Outreach and education: an example of good practice

The multidisciplinary Rett syndrome clinic held at St David's Hospital in Cardiff is an example of good practice in terms of outreach and staff exchanges. On occasion the whole team has gone to other hospitals including in Worcestershire and Devon, to hold a clinic. Individual team members have also visited Scotland to assist in education of staff there. Staff from other hospitals have been able to visit the clinics held in Cardiff, and this has resulted in St. Mary's Hospital in Manchester establishing its own service which will commence in March 2011.

## Current Situation

**It can be difficult both for patients and professionals to access reliable, up-to-date information on rare diseases and who the specialist(s) is/are in a particular condition.**

This lack of information can hinder the diagnostic process and also makes it difficult to signpost patients once a diagnosis has been made. Due to the rarity of these conditions, there is often a limited amount of reliable information available. For that reason it would be useful to have a place where all available information can be easily and readily accessed by professionals looking for further details on rare conditions. Some examples of good sources of information do exist, such as Orphanet<sup>18</sup>, the European portal for rare diseases and orphan drugs (see Recommendation 3a), but awareness of these resources is low so they are not being used to their full potential. There is a need to increase the profile of existing resources, and to increase coordination between resources to ensure that in-depth information can be accessed through one entry point, without duplication of existing work.

Improved access to good information would lead to empowerment of both professionals and patients. This would result in more informed decision making, which may ultimately have cost-saving implications for the

18. [www.orpha.net](http://www.orpha.net)



NHS as a result of fewer inappropriate tests, treatments or therapies being administered to patients. There may also be less wastage as a result of improved patient compliance. If patients are properly educated on any medication or treatments they are given, and they are able to develop a better understanding and knowledge of the benefits of such treatments, they are more likely to take their medication, resulting not only in less wastage for the NHS, but also improved patient outcomes.

### Recommendation 3

#### **Improve access to reliable information on rare diseases to make it easier for the public and professionals to obtain information.**

Improved access to reliable information will assist health professionals in making or rejecting a diagnosis. For the public, a source of reliable information would be invaluable both before and after diagnosis.

#### **Specific actions that need to be taken to improve access to information on rare diseases include:**

- a. An online portal sponsored by all UK health departments should be developed to provide links to reliable sources of information on rare diseases. This would include signposting current available resources, so rather than duplicating existing work, it would raise awareness of what is already available. The portal would not need to be elaborately designed, but rather would act as a point where information is laid out in a logical fashion to assist and direct professionals and patients. Links should be made to existing reliable information sources such as Orphanet, Dyscerne<sup>19</sup>, the Map of Medicine<sup>20</sup> and the UK Genetic Testing Network (UKGTN), as well as to all UK guidelines on rare disease that have appropriate accreditation. The portal need not be a costly project, however it would need to be managed and sustained to keep it up-to-date and relevant. It would be important for the portal to be available and utilised throughout the whole of the UK and recognised by patient groups and professionals alike.
- b. An assessment of information needed by healthcare professionals, and a review of existing resources, should be carried out to identify current gaps in this area. This should be coordinated by the Joint Committee on Medical Genetics on behalf of the Royal Colleges and the Deans of medical schools in the UK. An appraisal of this nature would take into account the resources that already exist, and would reduce the possibility of duplicating effort. It would help to identify which existing resources are frequently used and valued by healthcare professionals and which could be further developed or adapted to include other conditions. Following on from this appraisal, work should be commissioned to resolve the gaps in information needed by healthcare professionals.
- c. Orphanet is a European resource which aims to improve the medical care for rare diseases. It is supported by matched funding from the EU and the signatory countries (including the UK) as part of the Joint Action on Rare Diseases. It provides comprehensive information for professionals, patients, the public and industry. Orphanet UK is based at the University of Manchester and maintains a regularly updated information resource on UK rare disease clinical and diagnostic services, research programmes, and details of current clinical trials. As a Joint Action, matched funding is expected from the host country equivalent to the resource from the EU (to provide two information scientists in total in the UK). To ensure that this important service continues, and is comprehensive for the UK, we strongly recommend that the matched funding is provided at a level to ensure effective provision of information to professionals and patients, including where appropriate support for posts to deliver the functions of this service.
- d. Listed against each condition in the NHS Directory of Genetic Testing<sup>21</sup> should be a named clinician who can act as a source of advice and information for that particular condition. The role of providing information should be included as part of the named clinician's job plan. If a clinician was unsure whether a particular test was appropriate for a patient, they would be able to contact the named expert and seek their opinion. In December 2010 the UKGTN provided 688 genetic tests for 503 diseases; having access to information on each test would result in healthcare professionals being better able to request appropriate tests, which may lead to faster diagnosis of patients, and would also reduce NHS spending on multiple unnecessary genetic tests.
- e. Training on the use of diagnostic tools such as Dyscerne should also be provided to help healthcare professionals use them to their full potential. Investment has been put into developing good quality resources such as this, so they need to be utilised by the people they are aimed at. We recommend that

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19. [www.dyscerne.org](http://www.dyscerne.org)

20. [www.mapofmedicine.com](http://www.mapofmedicine.com)

21. [www.ukgt.nhs.uk/gtn/digitalAssets/0/767\\_DirectoryV5.pdf](http://www.ukgt.nhs.uk/gtn/digitalAssets/0/767_DirectoryV5.pdf)

development of a training package be included from the outset as part of such projects.

- f. An “index of suspicion” is required to provide guidance for doctors to ensure that they are referring patients to the appropriate specialist. This guidance should be created by the Joint Committee on Medical Genetics in collaboration with the Royal Colleges and medical schools in the UK. Development of an implementation plan would also need to be considered.

Improved linkage between specialist and local services (Recommendation 2) would also help professionals in local centres to access up-to-date information from specialist centres.

## Inequitable access to, and availability of, diagnostic tests

### Current Situation

**The criteria currently used by the National Screening Committee (NSC) to determine suitability for newborn screening tend to militate against rare conditions.**

The current criteria include considerations such as:

- There should be evidence from high-quality randomised controlled trials (RCTs) that the screening programme is effective in reducing mortality and morbidity.
- The clinical management of the condition should be optimised in all healthcare providers prior to participation in a screening programme.
- The condition should be an important public health problem.

While rare conditions are clearly an important health risk, their rarity makes high quality RCTs difficult to undertake. This should not be a barrier to progress for rare disease patient groups when compared with patients with a more prevalent condition for which clinical trials are easier to arrange. As generally understood and outlined in this report, the clinical management of rare conditions is not always equitable or well organised, and this is one of the reasons why families remain isolated and sometimes poorly served. While regrettable, rather than interpreting the lack of well organised services or carefully conducted treatment studies as a barrier to the introduction of screening, they should be viewed as deliverables that would result from a well organised national screening programme. Indeed, the organisation inherent in screening leads to a more careful scrutiny of service provision and treatment options.

The UK has been somewhat slower than other EU and North American counterparts to adopt the potential to screen for rare conditions. The outcome is that many children and families with these disorders in other countries are benefiting from prompt diagnosis and early treatment, which are denied to children born in the UK.

### Recommendation 4

**Appropriate rare diseases need to be considered for inclusion in the newborn screening programme.**

Other EU countries have modified their selection processes to ensure that rare conditions are treated more equitably when being considered for inclusion in newborn screening programmes. There is a need for the UK to consider a more targeted and timely approach for these disorders. We therefore recommend that rare diseases be considered for inclusion in the programme in the UK if they have a serious health impact on an individual, benefit from early intervention, and have a successful, approved treatment plan in place to adopt once diagnosed.

One such rare condition currently included in newborn screening programmes in the UK (excluding Wales) is medium chain acyl-CoA dehydrogenase deficiency (MCADD), thereby demonstrating that it is possible to include a rare condition that poses a high risk to an individual within a national newborn screening programme.

**Specific actions that need to be taken to more appropriately assess the inclusion of certain rare diseases into the newborn screening programme include:**



- a. There should be a critical re-appraisal of the current NSC criteria in relation to screening for rare conditions.
- b. The NSC should include a greater number of representatives with direct clinical experience of rare disorders when candidate conditions are being considered. The understanding of rare disorders and the benefits and risks of screening for these conditions is understandably limited and members may need to be co-opted when rare disorders are being discussed. Other countries have used relevant professional groups as a formal part of their assessment process and there is a need for the UK to do the same. The NSC should create a more formal mechanism to engage with a wide range of professionals to discuss conditions to be included in the newborn screening programme, in particular in terms of rare diseases where implications of the conditions may not be widely understood.
- c. The EU Newborn Screening Study that is currently being undertaken will be submitting recommendations on criteria for newborn screening in June 2011. This project is comparing practice in newborn screening programmes in all EU Member States in order to agree a set of recommendations that should help align practice. The UK is collaborating closely with this process and the study's forthcoming recommendations should be given serious consideration by bodies involved in screening in the UK.
- d. There is an opportunity for collaboration and partnership between private and public sectors to make it possible for conditions to be added to the newborn screening programme in a cost neutral way. The independent sector has a role to play in providing a high tech, cost-effective solution to the issue of expanding the newborn screening programme. There are valuable international examples of where such a partnership has been highly successful and cost-effective and this should be considered within the UK. In parallel, a closer integration and re-organisation of screening and specialist clinical services could result in significant overall cost savings to the NHS and provide a much improved service for some families with rare disorders. This approach should be considered by commissioners and planners of newborn screening programmes across the UK.

## Current Situation

### There is inequity of access to diagnostic tests across the UK.

This inequity can result in a more lengthy process of obtaining a diagnosis in some areas of the UK, meaning that some patients experience a delay in getting necessary treatment purely because of where they live.

Often access to diagnostic tests is inadequate because many specialised tests need to be referred on a provider to provider, cost per test basis to other centres within the NHS. This type of expenditure is an easy target for cost reduction and there is evidence that funds for these investigations are being restricted. However, this can result in examples of poor practice, such as hospitals referring patients to centres where the test is available in order to save the cost of testing. Although this ultimately results in the patient being able to receive the test, it does mean that the patient has to travel which may be difficult for them, so it is not a solution to the problem. Instead, mechanisms should be put in place for patient samples to travel, rather than the patient themselves. Patients may then travel to specialist centres if diagnosed with the condition, but should not need to do so to be tested for a disease that they may or may not have.

## Recommendation 5

### There must be improved access to diagnostic tests in under-served areas of the UK to ensure equity of access throughout the country.

#### Specific recommendations to improve access to diagnostic tests include:

- a. There should be a review of the architecture of testing for rare conditions in the UK. The NHS Commissioning Board in England, and the equivalent bodies in Wales, Scotland and Northern Ireland, should look at how testing is funded and delivered. The current system is inequitable and not fully effective and as such there must be a review, with follow-up work commissioned, to improve it. Appropriate diagnostic tests should be available to all patients where indicated, regardless of their location.
- b. There needs to be improved information on diagnostic testing available to healthcare professionals to ensure that they are only requesting appropriate, relevant tests for their patients. Many professionals are often unsure as to whether or not a particular test may be necessary, so guidance from an expert would be useful to help make assessments and decisions. This recommendation ties in with Recommendation 3d for a named clinician listed against each condition in the NHS Directory of Genetic Testing, and

Recommendation 3a for a portal to reliable information. The portal could have a page dedicated to information on diagnostic tests and their availability so that healthcare professionals can easily identify who best to contact for support. This approach would ultimately help to ensure more efficient use of NHS resources through better targeting of tests, and would also help with diagnosis of patients as healthcare professionals would be better informed on the conditions for which they are testing. Improved awareness of the UKGTN website and the information it provides should also be promoted. Furthermore, protocol-based investigative guidelines can improve equity and conserve resources by reducing unnecessary testing. Where appropriate, such guidelines should therefore be developed by professional groups seeking cost-effective solutions to good patient care.

### Benefits of a diagnostic service: an example of good practice

Primary Ciliary Dyskinesia (PCD) is a rare inherited condition, caused by ineffective ciliary clearance and affecting 1 in 15,000 Caucasians. Delayed diagnosis leads to bronchiectasis and inappropriate ENT surgery. Until 2006 there were problems with standardisation of, and inaccessibility to, diagnostic facilities for PCD in England. The patient support group and interested physicians worked together to secure a national service.

In 2006 the National Commissioning Group (NCG) funded a diagnostic service for England based at three collaborating centres (Southampton, Royal Brompton Hospital London and Leicester). Patients are referred for a diagnostic review, or nasal brushings are couriered from trained centres to ensure national coverage. The service includes:

- Three collaborating teams of doctors, nurse specialists, physiotherapists and scientists to provide and develop diagnostics for PCD.
- Nasal nitrous oxide measurement, high speed video, light and electron microscopy and cell culture to determine if disease is primary (inherited) or secondary.
- Standardisation of techniques and audit between centres.
- Training and advice on disease management to referring paediatric respiratory teams.
- Training in nasal/ bronchial sampling to referring courier centres.
- Physiotherapy advice and nurse specialist advice to families and to referring clinicians.

Referrals to the national service have increased annually, reaching 758 referrals in 2009-10, and approximately 15% of referrals are confirmed to have PCD. A national PCD database enables research and audit, which are informing improved management of this rare disease. A courier service ensures accessibility for all geographical regions.

This is an excellent example of a patient group successfully influencing the provision of services to lead to improved outcomes for patients, and it should be used as a model for other services.

### Recommendation 6

**Access to carrier tests for individuals and groups considered to be at significant risk of a specific condition should be facilitated and promoted.**

**Specific recommendations to improve access to relevant carrier tests include:**

- a. There are examples where local projects have been carried out that have identified at-risk groups in the local population. Where a need for carrier testing has been identified, processes should be put in place to ensure that testing is available and accessible to those who need it. Although establishing testing services may incur some cost, carrier testing would enable forward planning, which would result in savings to the NHS by enabling costly emergency procedures to be avoided. Carrier testing also provides prospective parents with reproductive choice in good time, and can facilitate timely diagnosis and effective management of any affected children. Once testing is set up, it may well be cost-neutral, or even cost-saving, as a result of avoiding expensive diagnostic measures and unforeseen interventions.

### Carrier testing clinics: an example of good practice

Guy's Hospital in London has a weekly walk-in clinic to test Ashkenazi Jews and their partners to identify whether they are carriers for Tay - Sachs disease, which has a much higher carrier rate among that population than the general population (a carrier frequency of about 1 in 25-30, as opposed to 1 in 250-300 ). This clinic has been successful in reaching the Ashkenazi Jewish population and Guy's laboratories now test about 700 people a year, a third of which come from the walk-in clinic. Programmes such as this should be used as a model for new initiatives in other conditions.

- b. Availability of carrier tests should take into account the frequency of the particular condition within the population, and should be available to people who are considered to be at high risk. For example, carrier testing for cystic fibrosis, which has a relatively high carrier frequency of 1 in 25 of the UK population, should be available to everyone within the UK, whereas testing for Gorlin syndrome, which has a carrier frequency of 1 in 40,000, would need to be for a specified population whose high risk had been identified. This would ensure that the NHS was only spending money on testing where it was likely to be most useful. The definition of 'at risk' therefore varies between conditions and must be taken into account when identifying who should be given access to carrier testing.
- c. Where services are in place to provide carrier testing, the sustainability of such projects needs to be considered. It is important that sustainability is a core component of commissioning and planning the service through effective workforce planning, thereby safeguarding the service from collapse when the interested individual retires or moves on.

# Commissioning and Planning

## Current situation

1. There is a wide variation in the health services available for patients with rare diseases across the UK.
2. There is often a lack of coordination between what is commissioned or planned centrally and what is commissioned or planned at the local level.
3. Provision of funding for specialised services for patients with rare diseases is perceived by some to be diverting resources away from local services.
4. Specialised services do not exist for every rare condition, and this can result in inequitable levels of care depending on which condition a patient has.
5. UK patients with rare diseases are being denied access to orphan medicines that have been granted European marketing authorisation.
6. There is a lack of information and guidance on the entitlements of patients who are refused funding for particular services, treatments or therapies.

## Recommendations

1. Commissioning and planning systems for rare diseases should ensure equitable access to health services and treatments across the UK, regardless of a patient's location.
2. Commissioning or planning structures should facilitate the coordination of what is commissioned or planned centrally and what is commissioned or planned at a local level.
3. The value of specialised services needs to be recognised and there should be resources safeguarded to fund these services.
4. Structures should be in place to ensure that patients are able to access the best care and support regardless of whether a specialised service exists for that condition.
5. There is an urgent need to reassess the mechanism and methodology by which the value of medicines for rare conditions is appraised for reimbursement on the NHS, to ensure improved and equitable access to licensed medicines from which patients will benefit.
6. Resources should be produced that inform patients of their rights and legal position if they are refused funding for treatments/therapies and how to go about the process of appealing a decision.

**England is currently undergoing a dramatic change in the structure of commissioning for healthcare services. We therefore hope that some of the current problems we describe in this section will be addressed by the new systems that will be in place by 2013. However, we feel that it is still important and of value to recognise what the current situations and challenges are, so as to provide a benchmark to which the new systems can be compared to identify successes and shortcomings of the new commissioning structure.**

**It is also important that these issues are recognised and addressed in Scotland, Wales and Northern Ireland where the new structure will not be relevant.**

## Wide variation in health services for rare disease patients across the UK

### Current situation

**There is a wide variation in the health services available for patients with rare diseases across the UK.**

Apart from those services that are commissioned or planned on a national level, variation in services for rare disease patients is often observed. This variation is due to the decisions taken at different levels of the commissioning or planning structure and results in patients having very different experiences, dependent on the condition they have and where they live.

Treatment and care is not of an equal quality for all patients with any particular condition throughout the UK. The 'postcode lottery' still exists whereby some patients are provided with a carefully planned programme of support and treatment (including drugs and other interventions where appropriate) and others are not, purely as a consequence of variations in the pattern of services planned and commissioned locally or regionally. Services differ within and between the home nations.

### Inequity of care: an example of bad practice in England

In England, the Specialised Services National Definitions Set (SSNDS) sets out definitions of 34 specialised services, each with a planning population of more than one million. The SSNDS exists to identify those services that are specialised and therefore should be subject to collaborative commissioning by Primary Care Trusts (PCTs) through the ten regional Specialised Commissioning Groups (SCGs). Many of the services outlined in the SSNDS will be accessed by patients with rare diseases. These arrangements have proved ineffective as currently none of the regional SCGs commissions all the services in the SSNDS and not one SCG commissions the same services as another.

If an SCG does not itself commission one of the definitions, it may still be commissioned in the region by PCTs. This could therefore lead to variation and a lack of coordination between different PCTs for those services that are not directly commissioned by an SCG, resulting in inequitable levels of care for a particular condition throughout the country. This situation is completely unacceptable.

PCTs across England take different approaches to commissioning specialised services. The lack of consistent, robust commissioning structures has led to confusion and a lack of transparency and communication about responsibilities in commissioning (or planning). This can lead to confusion and disagreements on funding, which could result in a delay in patients receiving the right service at the best time. Such a delay could have a detrimental effect on a patient's health and well-being.

Variations in the services available to patients with the same conditions lead to different health outcomes and quality of life, dependent on the patients' location. As an example of this, according to research by the Muscular Dystrophy Campaign<sup>22</sup>, poor service provision for patients with Duchenne muscular dystrophy in the south west of England led to an average age of death for patients of 19 years, compared to 30 years in the north east of England (before all services were reviewed).

22. 'Building on the Foundations: The need for a specialist neuromuscular service across England', Muscular Dystrophy Campaign, December 2007

## The 'postcode lottery': examples of bad practice

In response to our consultation on our initial recommendations, RDUK was informed of a number of examples of bad practice showing the existence of a postcode lottery for access to treatments or services.

The ChILD Lung Foundation UK gave the example of inequality of access to palivizumab injections. This injection is given to affected children under the age of two to protect against the respiratory syncytial virus over the winter months. ChILD told us that they were aware of a family in Northern Ireland that had been able to access this injection, whereas at a similar time a child in Scotland was denied access, despite having very similar symptoms and oxygen requirements. The child in Scotland had to be hospitalised three times over the winter months, which may have been preventable had the injection been made available. Emergency admission to hospital would also probably have been more expensive than administration of the drug.

The PSP (Progressive Supranuclear Palsy) Association told us that even within the five Trust regions of Northern Ireland there is a huge discrepancy in the provision of care to patients with PSP and cortico basal degeneration (CBD). Care packages there can range from 'excellent to appalling' and budgets 'seem to vary greatly between the Trusts resulting in great inequity within Northern Ireland alone'.

The Association for Spina Bifida and Hydrocephalus (ASBAH) also told us that patients are subject to the postcode lottery. ASBAH reported that some authorities provide district nurses to attend to dress patients' wounds, whereas in neighbouring authorities patients have to dress their own wounds or attend a clinic on a specified day to receive assistance.

These examples are just a few of the many we have heard, and prove that the postcode lottery does exist within the UK, and there is an urgent need for it to be addressed.

There is also the issue of patients in one home nation not being able to access a service or treatment that is readily available and accessed in another home nation. Patients often report experiencing difficulties in accessing professionals that are based in a different home nation to themselves, which again contributes to an inequity in the services received. Although this is not an issue for all rare disease patients in the UK, it is an issue of principle that needs to be addressed to ensure that all rare disease patients within the UK are able to access services that would be of benefit to them, regardless of the home nation in which they live and in which the service is provided.

### Recommendation 1

**Commissioning and planning systems for rare diseases should ensure equitable access to health services and treatments across the UK, regardless of a patient's location.**

**Specific actions that should be taken to achieve equitable access to health services and treatments include:**

- a. It is vital that the commissioning and planning of specialised services for patients with rare diseases are carried out at the appropriate planning population level to avoid unnecessary and inequitable variations and to ensure that the service is developed. The specialised commissioning bodies of the UK should work to ensure that commissioning or planning at the appropriate level is achieved. It is hoped that the new structure in England will work to ensure that this is the case, but it is important that this occurs within all devolved nations.
- b. There should be a review into the current structures and policies for accessing health services across the home nations, and the restrictions that prevent access. If a service exists in one nation, we believe that it should be accessible by all UK residents who need it. A review of the bureaucracy surrounding access between the home nations would ensure that there were no unnecessary barriers in place blocking access to good quality services needed by patients.
- c. There is a need for the specialised commissioning and planning bodies in each of the home nations to work together actively to improve accessibility to services for rare diseases, as due to the low numbers of patients affected by individual diseases, it is clearly not possible for services to be established for all rare diseases in each of the four home nations.



Currently the devolved nations have observer status on the Advisory Group for National Specialised Services (AGNSS) in England. Scotland, Wales and Northern Ireland are also invited to participate in regular Specialised Commissioning Group Directors' meetings. We recommend that this relationship between home nations is developed as the NHS Commissioning Board in England is introduced, and that more formal links are developed to assist communication between all UK commissioning boards. This would assist collaboration between home nations to help ensure accessibility of the specialised services in existence in the different parts of the UK.

## Lack of coordination between central and local commissioning and planning

### Current situation

**There is often a lack of coordination between what is commissioned or planned centrally and what is commissioned or planned at the local level.**

Not all services required by a patient with a rare disease are specialised. Indeed, the majority of services needed by a patient on a day-to-day basis will be non-specialised services commissioned and planned at a local level. A lack of integration between services commissioned or planned at different levels results in inefficient collaboration and coordination, which can mean that patients do not receive the optimum level of care. A patient may receive a good quality service commissioned or planned nationally, for example, but can then struggle to access good quality services they need in their local community. There is therefore the need for commissioners and planners to ensure better integration between these services to ensure that patients are able to access the treatment and care needed, and to avoid conflicting messages between professionals working locally and those in specialised centres.

### Recommendation 2

**Commissioning or planning structures should facilitate the coordination of what is commissioned or planned centrally and what is commissioned or planned at a local level.**

As the majority of a patient's care will be provided at a local level, there must be good coordination and communication between commissioning and planning bodies to ensure integration and a thorough understanding of each other's roles and how services are linked.

**Specific actions that should be taken to achieve integration and linkage between commissioning and planning bodies include:**

- a. A strong oversight body is necessary to ensure this integration and linkage. Consideration of the integration of services at multiple levels should become a mandatory aspect in the commissioning and planning of new services. The new NHS Commissioning Board is in a strong position to ensure this integration in England. In Wales it should be the responsibility of the Welsh Health Specialised Services Committee, in Scotland of the National Services Division, and of the Health and Social Care Board in Northern Ireland.
- b. There must be comprehensive guidelines agreed, developed and implemented by commissioning and planning bodies, in consultation with patients and healthcare professionals, to ensure that it is clear and transparent which component of a patient's care is the responsibility of which body and that services are integrated across the whole care pathway. There need to be systems in place to create a relationship between all levels of commissioning and planning, to ensure that service provision is seamless.
- c. Commissioners and planners must take into consideration that the distribution of patients will not always be even, due to the greater prevalence of some rare diseases in certain populations, e.g. some conditions are more common in population sub-groups. Examples include thalassaemia in Mediterranean populations, and the increased prevalence of recessive single gene genetic conditions in people of Pakistani origin. Effective commissioning and planning structures should be able to take account of such local needs.



## Perception of funding for specialised services

### Current situation

**Provision of funding for specialised services for patients with rare diseases is perceived by some to be diverting resources away from local services.**

Specialised services for rare diseases ensure that patients have access to the services and expertise they need, planned and commissioned at the appropriate population level. This should not be seen as a distraction from the day-to-day business of providing healthcare to the 'average' citizen. Specialised services should be considered complementary to local services as opposed to a threat to local services, and commissioners should be in a position to facilitate coordination between these areas.

Evidence for the necessity of commissioning at national level is amply provided by the progress made by the National Specialised Commissioning Team in safeguarding services for some of those with very rare conditions. Commissioning of services such as these would be impossible at a local or even regional level.

### Convoluting funding systems: an example of bad practice in England

Currently, funds in the English NHS follow a convoluted pathway. The budget is top-sliced to provide funds for national commissioning. It is subsequently distributed between PCTs, who then top-slice the funds again for Strategic Healthcare Authority level planning (Regional Commissioning) and use the remainder for commissioning of services within their area of remit. This funding method, whereby funds for specialised services appear on PCTs' balance sheets, is an important cause of PCT hostility towards specialised services, in the opinion of RDUK.

### Recommendation 3

**The value of specialised services needs to be recognised and there should be resources safeguarded to fund these services.**

**Specific actions that should be taken to ensure recognition of the value of specialised services and safeguarding of resources to fund them include:**

- a. Budgets for funding specialised services should be protected and allocated specifically to the specialised commissioning or planning body. This would ensure that in times of financial pressure, specialised services would not be unfairly targeted due to the above perception and rhetoric of localism. The budget assigned for specialised services should be flexible and allow expansion when evidence suggests that services would be better commissioned/planned by the specialised commissioning or planning body. If a service formerly commissioned or planned at a local level becomes commissioned or planned at a national level, then the budget for the delivery of that service should be allocated accordingly.

## Inequity of care between rare conditions

### Current situation

**Specialised services do not exist for every rare condition, and this can result in inequitable levels of care depending on which condition a patient has.**

It is widely acknowledged that patients who have access to a specialised service for their rare condition have better outcomes in terms of both their health and general well-being than those who do not. As there are no specialised services available for many rare conditions, there are inequitable levels of access to good quality services depending on the condition a patient has.

Specialised services for rare conditions commissioned at a national level in England have tended to be introduced in response to ad hoc applications from service providers. While we support the AGNSS' new decision making framework, which increases the transparency and rigour applied to the decision making process, consideration should be given to ensure a balance between the need for evidence-based services against the burden placed on applicants, which may include clinicians working alongside patient groups.

Many rare disease patient organisations are entirely volunteer-led and under-resourced, so they may not be able to proactively drive forward applications for national commissioning. There are also many conditions

for which there are no patient organisations, which may mean that these conditions are less likely get a nationally designated service. While we believe that involving patient organisations in the commissioning process is important, it should not be to the detriment of those conditions with less well-resourced patient organisations, or those with no patient organisation to take things forward.

The current reactive approach to commissioning specialised services for very rare conditions is unsustainable and leads to the danger of services being commissioned only for those who 'shout the loudest'.

#### **Recommendation 4**

**Structures should be in place to ensure that patients are able to access the best care and support regardless of whether a specialised service exists for that condition.**

We recognise that some rare conditions affect such a small number of patients, or are insufficiently well understood given current knowledge, that it would be challenging to designate a specific specialised service, especially as there are over 6,000 rare diseases. It would also be impossible to commission and plan an individual specialised service for each specific rare disease. However, we believe that even for those conditions that cannot feasibly have a specific specialised service, structures should be in place to ensure that patients are still able to access good quality services.

**Specific actions that should be taken to achieve better development of specialised services include:**

- a. Specialised services should be commissioned and planned for clusters of conditions with similar needs. This would alleviate the problem of not being able to commission and plan separate services for each rare disease, and it would lead to a more proactive approach to commissioning. We recognise that the SSNDS makes use of groupings of conditions, and we believe that this approach could be applied to conditions affecting smaller patient populations. This approach would also benefit those rare diseases that lack well-resourced patient organisations to drive forward applications. These centres of excellence for clusters of conditions are discussed further in Recommendation 1 of the Delivering Coordinated Care section of this report. The new NHS Commissioning Board in England should undertake a pilot project looking at how services based around clusters of conditions can be best achieved with the aim of expanding it more widely for other groups of conditions. All home nations should participate and be involved in this initiative.
- b. Following the pilot project discussed above in Recommendation 4a, an appraisal should be carried out of which conditions need to be prioritised to receive specialised services based on the clinical need of the UK, and the relevant bodies should be empowered and resourced to do this.
- c. Models of good practice in commissioning or planning services for rare diseases should be shared so that these can be expanded on for other conditions. The effective services that currently exist should serve as a model for developing new services.
- d. Patient organisations and clinicians should be supported adequately by commissioning and planning bodies to understand and fulfil the requirements of the application process for specialised services.
- e. The new NHS Commissioning Board in England and equivalent specialised commissioning bodies in Wales, Scotland and Northern Ireland, should periodically review the services for which they are responsible, to ensure that the services are achieving their aims and are still relevant for national commissioning and accessible to all who require them. This process would ensure that services are working effectively and efficiently, and could also take innovation into account.

## Access to orphan medicines

### Current situation

**UK patients with rare diseases are being denied access to orphan medicines that have been granted European marketing authorisation.**

In the UK, there are no consistent funding routes for orphan medicines. In England, many orphan medicines are not evaluated through the health technology appraisal (HTA) process and the few that are appraised are often rejected on the basis of their high estimated cost per quality-adjusted life year (QALY). In the absence of NICE guidance, decisions on whether or not to fund treatments are often being made by PCT individual funding request (IFR) panels, groups with little experience of the specific issues surrounding the appraisal of orphan medicines. Not only does this lead to duplication of effort, with over 150 bodies making funding decisions on the same medicines, it also inevitably leads to inconsistency in the decisions being made. This in turn leads to inequity of access to the medicines being considered.

In some areas collaborative decisions are made about which medicines to fund and we hope that the new commissioning system in England will build on this expertise.

By contrast, in Scotland, where the Scottish Medicines Consortium (SMC) appraises all new medicines coming to market, as of May 2010 the SMC had appraised 46 orphan medicines, recommending 18, rejecting 17 and recommending the restricted use only of a further 11. This situation has come about despite the addition of modifiers to the SMC process designed to give special consideration to treatments for rare disease and terminal illness.

In Wales, the All Wales Medicines Strategy Group (the appraisal committee) has a policy for the special consideration of ultra orphan medicines (UK prevalence of 1:50,000), but not for orphan medicines.

### Recommendation 5

**There is an urgent need to reassess the mechanism and methodology by which the value of medicines for rare conditions is appraised for reimbursement on the NHS, to ensure improved and equitable access to licensed medicines from which patients will benefit.**

**Specific recommendations to ensure equitable access to licensed medicines include:**

- a. Orphan medicines should be subject to evaluation, but methods and processes should be refined for orphan medicines to take into account the difficulty of collecting data for small populations as well as the costs associated with developing drugs for small populations. Current HTA methods and processes, and the cost effectiveness thresholds that are applied as part of them, may not always be appropriate for evaluating orphan medicines. Although NICE HTA processes have evolved considerably, there remain concerns around how NICE takes into account the specific factors that are important for appraising orphan medicines, such as rarity and innovation.

Evaluation should be based on an appraisal of the technology against multiple criteria and not simply a cost utility analysis. A recent positive example of this approach is the decision making framework developed by the AGNSS. This framework involves a consideration of 'value' in four domains: health gain, societal value, reasonable costs and good practice. This approach is currently undergoing pilot testing and we look forward to reviewing its outcome. Unfortunately, however, the use of this mechanism will be reserved for medicines that are used to treat 500 or fewer patients.

- b. Funding for specialised drugs/treatments/interventions for rare diseases should be organised nationally from a central source, to avoid inconsistencies in access and duplication of resources and to ensure that decisions are made at the level where the best expertise is available. This is particularly important for treatments that have not been appraised by NICE.
- c. At the end of the current Pharmaceutical Price Regulation Scheme, which expires at the end of 2013, the UK government intends to move to a system of Value Based Pricing (VBP). The stated purpose of VBP is to improve NHS patients' access to effective and innovative drugs by ensuring that they are available at a price that reflects the value they bring. As the detail of VBP is worked out, it is imperative that the issues specific to the appraisal of orphan drugs are fully considered. It is also important that patient input is included as part of the decision making process.

## Lack of information on patients' entitlements when funding is refused

### Current situation

**There is a lack of information and guidance on the entitlements of patients who are refused funding for particular services, treatments or therapies.**

Many patient organisations do provide guidance to their members; however, we are concerned that there are no patient organisations for many rare diseases, leading to the situation whereby those who 'shout the loudest' can appeal successfully, and those who are not supported by a patient group can be left in the dark.

When the health service undergoes reorganisation, systems and groups change, which can have a detrimental impact on a patient's understanding of their rights and how to negotiate their way through the system. In England, given the changes that are currently taking place within the NHS, it is even more important that patients know what they can expect to receive from the NHS Commissioning Board and from GP Commissioning Consortia. It is vital that the distinction is made as transparent as possible to ensure understanding among patients. The process by which a patient can appeal a funding decision must also be made clear and transparent.

Likewise, with the recent reorganisation of NHS Wales abolishing the internal market, information and resources should be readily available to assist and inform patients of the role of the Health Boards and associated groups in relation to planning and delivering their health services.

Even as new structures settle into place, it is vital that patients continue to be aware of their rights and how to challenge decisions. This applies across all home nations and will always continue to do so.

### Recommendation 6

**Resources should be produced that inform patients of their rights and legal position if they are refused funding for treatments/therapies and how to go about the process of appealing a decision.**

These guidelines could then be further developed by patient organisations to make them more specific to their condition, where this is relevant.

The NHS Constitution for England states that the NHS commits to 'provide you with the information you need to influence and scrutinise the planning and delivery of NHS services'<sup>23</sup>, and it also commits to 'make decisions in a clear and transparent way, so that patients and the public can understand how services are planned and delivered'<sup>24</sup>. It is therefore vital that relevant information and guidelines are produced to uphold this commitment. It should be part of the NHS Commissioning Board's responsibility to establish who should take this role forward. It may be a role for HealthWatch in England.

The Welsh Assembly Government has announced that it is developing an Independent Appeals Model for Individual Patient Commissioning and the Community Health Councils are charged as the NHS Wales 'watchdog'. We recommend that the Community Health Councils be responsible for developing these guidelines for patients in Wales.

In Scotland, we recommend that the responsibility should lie with the Independent Advice and Support Services and Health Rights Information Scotland, and in Northern Ireland with the Patient and Client Council.

23. 'The NHS Constitution for England', Department of Health, January 2009, page 7 [www.dh.gov.uk/prod\\_consum\\_dh/groups/dh\\_digitalassets/documents/digitalasset/dh\\_093442.pdf](http://www.dh.gov.uk/prod_consum_dh/groups/dh_digitalassets/documents/digitalasset/dh_093442.pdf)

24. 'The NHS Constitution for England', Department of Health, January 2009, page 5, as above

# Patient Care, Information and Support

## Current Situation

1. Patients are not provided with sufficient reliable information on their medical, psychological, social and other needs at diagnosis and throughout the progression of their condition.
2. Information for patients is not always provided in the appropriate format and at a level that ensures that the patient will be able to understand and use the content to support effective and appropriate decision making.
3. Patients frequently report not being offered psychological support in relation to their condition.
4. Links between the medical and social aspects of care and support for patients with rare diseases are often weak and support for non-medical needs is severely lacking.

## Recommendations

1. Patients should be provided with ongoing, reliable information on their condition and how to manage it, including any existing treatment options, and how to receive the support they need.
2. Information should be made available in various formats and at various levels of scientific and medical knowledge.
3. Psychological support for the whole family should be considered an integral part of the care package.
4. Social support for those affected by rare diseases should be a fundamental part of the patient's care package.

## Lack of information provided to patients with rare diseases

### Current situation

**Patients are not provided with sufficient reliable information on their medical, psychological, social and other needs at diagnosis and throughout the progression of their condition.**

This lack of information may result in:

- feelings of isolation
- loss of faith in the healthcare system
- uninformed decision making which can lead to the patient or family mismanaging their condition, potentially resulting in the deterioration of the individual's health; sometimes more expensive interventions are subsequently needed or patients are dealt with by A&E in crisis.

Too often patients are given a diagnosis but no further information, and are left to research their condition on their own. This frequently results in patients finding unreliable, often alarming information and not being able to discuss this with anyone who understands the condition. RDUK's recent survey of patient experiences found that over half of patients (52%) feel that they are not given enough information on their condition on diagnosis. In examples of particularly bad practice some patients are given no information at



all apart from the name of their condition. In response to RDUK's recent survey, many patients reported that they came across information only by chance, such as an article in a newspaper, after years of living with a condition.

A patient's condition may be being managed effectively by the NHS according to best current knowledge on that condition, yet the lack of appropriate people and/or tools to communicate that information, together with a patient's feeling of insecurity or isolation, can lead to a perception of inadequate care. Information is a crucial element of the service a patient receives and should not be viewed in isolation or as something which is optional. Empowering patients through information is a relatively low-cost way of ensuring better management of a condition as well as increasing a patient's satisfaction with the service they are receiving.

As information on rare diseases and their management is often more scarce and difficult to find than information on common diseases, it is of even greater importance that patients with rare diseases are supported by the NHS with the information they need.

## Recommendation 1

**Patients should be provided with ongoing, reliable information on their condition and how to manage it, including any existing treatment options, and how to receive the support they need.**

**Specific actions to improve the information patients are given include:**

- a. An 'information prescription' should be given to all patients when they are first diagnosed with a rare condition. Such as approach is in line with the current ethos central to the health reform in England, "No decision about me without me", although it is equally valid for patients throughout the UK. The prescription should outline the information patients and families have identified as important, and define the way they should receive it, when they should receive it and from whom. A copy of the prescription should also be sent to the patient's GP so that the primary care physician is kept informed and can use the resource to find out more about the condition in order to better support their patient

We recommend that a generic outline information prescription be developed that can then be tailored to each patient regardless of their condition. Headings within this document could include:

- medical information, including where appropriate, possible treatment options
- when the patient will receive their care plan
- information needed by carers/family
- social information
- financial information
- educational information
- welfare information
- palliative care information
- how to access information on research into the condition
- any other relevant information or sources of further information.

The information prescription should outline the different professionals available to offer the various types of support including, where necessary, social services and psychological support professionals. Contact details should also be provided. This would enable patients to receive comprehensive information to help guide them after diagnosis. They can then see what information they should expect to receive during the management of their condition, at an appropriate pace that they could regulate to best suit them. It would also allow patients to see what support they are entitled to, and to contact the appropriate person or provider if they do not receive it.

An information prescription for all patients with a rare disease would enable patients to feel assured that information will be ongoing, and that, rather than just receiving a name and website reference, there is an action plan in place detailing when, how and what information they will receive.

Details of how to access information about research into a patient's condition should also be included in the information prescription. England's NHS constitution states that: 'The NHS will do all it can to ensure that patients, from every part of England, are made aware of research that is of particular relevance to them.'<sup>25</sup> This should not only be the case for patients with common diseases, but for those with rare diseases as well, and it should be extended to all the home nations, yet it is a commitment which is not always implemented, even in England.

As there are no effective treatments for most rare diseases, information on new research is often particularly important to many families. Encouraging patients to join a registry, where one exists, would for example help to alert patients to clinical trials for which they may be eligible. This is supported by Recommendation 3a in the Coordination of Research section of this report.

An information prescription is already in development for cancer patients, including those with rare cancers, in line with the Cancer Reform Strategy<sup>26</sup> that is being developed by the DH with input from patient organisations. We suggest that this be used as a guide for the development of an information prescription for other rare diseases in addition to rare (and common) cancers, to avoid repetition of existing work. A rare disease information prescription should be developed by the health departments as a pilot project for a particular condition(s) in collaboration with patient organisations, in order to assess its effectiveness and to identify what details should be included and how such an information prescription would best work for rare diseases. Following the pilot, the information prescription should then be developed and rolled out to cover all rare diseases.

- b. On diagnosis patients and families need to be made aware of available reliable sources of information that they may want to use to research their condition or access support and information. These sources should include the relevant patient organisation/s where they exist. Patient organisations often have forums and helplines where patients are able to discuss their concerns with others who have experienced similar situations. This is a simple, effective way to address some of the support needs that patients may have.

This recommendation ties in with that for a web portal sponsored by the health departments that would link to reliable online information to assist with the prevention and diagnosis of rare conditions (Prevention and Diagnosis, Recommendation 3a). If this portal were put in place, healthcare professionals would know where to access information themselves and have a starting point to which they could also easily direct patients wanting to do their own research, and be sure that it was reliable source of information.

We acknowledge that online informal social networking groups exist for rare diseases. While we recognise that these groups can assist communications between patients, carers and families, people using any medical information discussed on these group sites should be made aware that this information may not necessarily be accurate or have been accredited or endorsed by medical professionals.

- c. Patients should be offered a face-to-face appointment with a relevant healthcare professional at a designated time after initial diagnosis to discuss their next steps and queries. The interval to this appointment could depend on the nature of the condition, but our research suggested that four to six weeks after initial diagnosis would be an appropriate time for this meeting. This would allow time for the patient and family to digest the diagnosis and think of any questions they may have for discussion with an experienced professional. The production of 'Frequently Asked Questions' sheets would also be helpful to encourage patients and their families to think about relevant questions that they may have.

Some rare cancer patients currently have such a follow up appointment several days after diagnosis and it has been shown to be an effective way of delivering information to patients. Rare cancer patients need an immediate appointment with a healthcare professional and often commencement of treatment on a fairly urgent basis, therefore information is needed immediately in order to make informed choices about treatment and support. The format of this appointment should be adopted as a model of good practice for all rare disease patients.

25. 'The Handbook to the NHS Constitution', Department of Health, March 2010 [www.nhs.uk/choiceintheNHS/Rightsandpledges/NHSConstitution/Documents/COI\\_NHSConstitutionWEB2010.pdf](http://www.nhs.uk/choiceintheNHS/Rightsandpledges/NHSConstitution/Documents/COI_NHSConstitutionWEB2010.pdf)

26. 'Cancer Reform Strategy, Chapter 5 – Living with and Beyond Cancer', Department of Health, December 2007 [www.dh.gov.uk/prod\\_consum\\_dh/groups/dh\\_digitalassets/documents/digitalasset/dh\\_081007.pdf](http://www.dh.gov.uk/prod_consum_dh/groups/dh_digitalassets/documents/digitalasset/dh_081007.pdf)



The type of professional involved in this meeting may vary between conditions and patients, but needs to be someone with experience or knowledge of the condition and of the implications of it, and someone who is able to communicate well with the patient on a level appropriate for them. The person best placed for this might be the Care Coordinator (see Recommendation 3b, Delivering Coordinated Care). In the case of rare cancers, and where a patient is fortunate enough to have a clinical nurse specialist (CNS), we believe that this is an appropriate person to deliver this service.

For many rare conditions there may only be one experienced professional or specialist in the country. Where this is the case, there should be better linkage between that person and the local service in order to enable education of a professional within the patient's local centre. This ties in with the recommendation for improved linkage between specialist and local services (Recommendation 2, Prevention and Diagnosis). It may also be appropriate to facilitate a meeting between the specialist and the patient, should the patient wish to liaise directly with this person. A face-to-face meeting should always be offered, but if this involves travelling and the patient/family would prefer not to travel for their information appointment, other possibilities such as video-link should be explored.

- d. Every NHS care provider, such as NHS Trusts and Health Boards across the UK, should have a referral facility to assist patients and families in accessing information. Although online access to information about rare diseases is becoming increasingly important and widespread, there are still some families who may not have access to a computer and the Internet at home, so such a service would be of particular benefit to them.

Patient Advice and Liaison Services (PALS) exist within England and are generally situated in hospitals. This is a model that could be replicated and rolled out in other home nations. Although the concept of PALS is good, we recommend that it be improved to reach its full potential. There is a need for these services to be given the most relevant and up-to-date resources so that they are in a strong position to offer high quality information to patients and families. This should include resources such as an up-to-date list of relevant patient support groups and an up-to-date 'Contact a Family directory - The essential guide to medical conditions, disabilities and support'<sup>27</sup>. The basic infrastructure of the PALS service is already in place, and as such could be easily developed and improved to make it more effective, without incurring a substantial cost. Ideally these services should provide a designated computer and a knowledgeable assistant to help patients and families research their condition. The assistant would be able to give guidance on relevant, reliable websites, and be on hand to assist those who are not computer literate.

- e. Healthcare professionals should ensure that patients are given information on all available and appropriate treatments at various stages of their condition. While we recognise that for many rare diseases there is no specific treatment available for patients, for those conditions that do have treatments, patients should be made aware of them. There is evidence that patients want to be given choice around treatment options, and so there is a need to support them in doing so.
- f. In order to be better supported and empowered, patients need a designated person to whom they can go with questions. This person may be the Care Coordinator discussed in the Delivering Coordinated Care section of this report.

Currently this service is often provided by patient organisations. Where this is the case we recommend that government funding is made available for patient organisations to provide this role in a sustainable manner. The opportunity exists for an improved partnership between the health departments and patient organisations in the provision of this service. Patient organisations have the skill and expertise to fulfil this role effectively, but are often lacking the financial resources to be able to do so.

Joint funding from the health departments in the UK would allow patient organisations to optimise their skills in this area and to fulfil this necessary role. There is evidence from current examples of good practice that health departments have already recognised that many patient organisations are often successfully carrying out this role. Existing good practice should be expanded to enable services to continue/ be established rather than solely relying on third sector funding, which is not always adequate or sustainable.

It is to the detriment of patients and the NHS to allow a service which has been shown to be necessary and effective fall by the wayside as a result of lack of funding, As such there should be consideration of sustainability of these services and it should be recognised that patient organisations can help the NHS to provide good quality services to patients.

27. [www.cafamily.org.uk/medicalinformation/conditions/aboutthedirectory.html](http://www.cafamily.org.uk/medicalinformation/conditions/aboutthedirectory.html)

## Partnership between charities and health departments: examples of good practice

An example of this type of partnership exists with muscular dystrophy regional care advisors. These advisors offer practical and emotional support for people with muscle disease and their families. These positions were originally fully funded by the Muscular Dystrophy Campaign (MDC), are currently funded 50:50 by the MDC and the regional Specialised Commissioning Groups in England (SCGs), and will be fully funded by the regional SCGs from April 2011. Funding from the NHS has enabled the MDC to employ more care advisors so that families elsewhere in the UK are able to benefit from the invaluable support they offer. The MDC have found that 'investment in these posts is cost-effective as they have been shown to save consultants' time, reduce emergency admissions and re-admissions, reduce hospital stays and coordinate care locally'<sup>1</sup>. We recommend that this model is rolled out for other rare conditions.

The Huntington's Disease (HD) Association provide regional care advisors whose role includes giving information and advice to families affected by HD in England and Wales, answering crisis calls, liaising with professional service providers, and providing awareness-raising workshops. This is an example of one of the services that would benefit from some Department of Health funding to ensure continuity and sustainability of service and to make sure that the service is sufficient to meet the population's need.

One example of where sustainable funding is needed is with regard to the Rarer Cancers Foundation (RCF) patient helpline, which provides effective support to those diagnosed with these diseases. The RCF was given three years' funding for this project, which has been incredibly helpful and well received by patients and families. Financial support for this initiative ends in 2011 and unless further funding can be found, the service will have to be significantly reduced. This will have a direct negative impact on the patients and families who use it.

<sup>1</sup>. 'Access To Specialist Neuromuscular Care: The Walton Report.', All Party Parliamentary Group for Muscular Dystrophy

## Information is not always provided in the most appropriate format for patients

### Current situation

**Information for patients is not always provided in the appropriate format and at a level that ensures that the patient will be able to understand and use the content to support effective and appropriate decision making.**

Patients often report that they were not given the level and amount of information they would have liked, or that they did not understand all of the information given to them. Patient organisations often do provide good quality information, but there is no patient organisation in the UK that is dedicated to supporting those affected by many specific rare diseases. There are also occasions where access to good quality information produced by patient organisations has been blocked by unnecessary bureaucracy, for example for not having the NHS Trust's logo on it.

### Recommendation 2

**Information should be made available in various formats and at various levels of scientific and medical knowledge.**

**Specific actions that should be implemented to improve patients' ability to understand and fully utilise information include:**

- a. Good quality information is often produced by patient organisations, but where there are gaps in the information available on certain conditions funding should be provided to produce this information. This should be done by health departments in collaboration with patient organisations. Empowering patients by giving them information about their condition and how best to manage it is a cost-effective way of ensuring better outcomes for patients.

Children Living with Inherited Metabolic Diseases (Climb), the charity for those affected by metabolic diseases, has recently produced a series of DVDs for patients, thanks to a Department of Health grant<sup>28</sup>. We recommend that similar grants be provided to other patient organisations to assist them in developing their own information material.

- b. Information should be made available in a variety of formats including leaflets, journals, websites and DVDs. Information should be patient-specific, in plain language, and developed to a level and format that best suits them. Information should be written in a way that promotes effective interpretation and use, and patients should be supported to ensure they understand all the information they are given. The type of information required will vary between patients. Some will want all available information on their condition, including the science and basis for the disease, whereas others may want much less detail. We therefore recommend that information should be produced at various levels of scientific and medical knowledge.

As an example of good practice, the Cystic Fibrosis Trust has recently produced a video aimed at educating children with cystic fibrosis<sup>29</sup>. This has been well received by parents to help them share information with their children.

- c. Links to good quality information should be provided by the portal recommended in the Prevention and Diagnosis section of this report, Recommendation 3a. Such links will allow patients and families to find information more easily and to select the level of information that suits them. While we are aware of the importance of accrediting information, methods of accrediting information to be included on the portal should not be onerous for patient organisations. Many patient organisations for rare diseases are small or run entirely by volunteers, and so do not have the resources to go through protracted processes of accreditation.
- d. When good quality information has been developed by a patient organisation, unnecessary bureaucracy should not restrict the availability of this information. If a healthcare professional thinks that a source of information would be useful to a patient, NHS healthcare providers should work alongside them to make this information accessible.

As an example of bad practice, Unique, the rare chromosome support group, has been told by a clinical geneticist that his NHS Trust prohibits him from giving his patients copies of any information that does not carry the Trust's own logo. This includes even high quality information guides that have been verified by medical experts, such as those produced by Unique on rare chromosome disorders. The geneticist instead has to direct families to the Unique website, which means that families without access to the Internet miss out on the information, even though the geneticist has judged it to be suitable.

## Lack of support for rare disease patients' non-medical needs

### Current situation

#### **Patients frequently report not being offered psychological support in relation to their condition.**

RDUK has found that 71% of patients do not feel they receive sufficient psychological support in relation to their condition. Patients could often benefit from professional support in this area, particularly at transition times and other important periods of their life. Carers also need psychological support. They may feel overwhelmed at times, and need essential support to provide the best care they can. Caring for a loved one can be very difficult and the impact of this on the carers is not always recognised and addressed.

Too often this support is not given, and families must exert pressure to get the help they need. This should not have to be the case.

28. [www.climb.org.uk/bemis\\_project.htm](http://www.climb.org.uk/bemis_project.htm)

29. [www.youtube.com/watch?v=Wu172eMrlQI](https://www.youtube.com/watch?v=Wu172eMrlQI)

### Recommendation 3

**Psychological support for the whole family should be considered an integral part of the care package.**

**Specific actions that should be taken include:**

- a. The delivery of psychological and emotional support for both the patient and their family or carer should be considered from the outset of the patient's care and should be included as part of a patient's care plan. Feelings of isolation and fear are often more extreme for patients with these conditions due to their rarity, and so the need for psychological support is often great. Those with more severe distress should be given access to counsellors, psychologists and psychiatrists.
- b. Making patients aware of the various sources of support available to them and encouraging them to make use of this support whenever necessary. Sources of support should be included in the information prescription (Recommendation 1a). Recommendation 1d (an advice service in hospitals) would also help to direct patients to sources of support.
- c. The provision of a Care Coordinator, as recommended in Delivering Coordinated Care, Recommendation 3b, would help by providing support to patients as well as reducing the sense of isolation rare disease patients often feel by providing a point of contact for questions and advice about their care and condition. RDUK found that 37% of patients with rare diseases do not have anyone that they can contact with questions about their condition.

### Current situation

**Links between the medical and social aspects of care and support for patients with rare diseases are often weak and support for non-medical needs is severely lacking.**

RDUK's recent survey of patients' and families' experiences found that only 33% of patients reported receiving adequate support with their social needs and only 29% with their financial needs. Benefits, such as the Disability Living Allowance (DLA), Carer's Allowance and Employment and Support Allowance, may be available to financially assist patients with rare diseases and their families, but too frequently patients are not aware of or informed about these, and are not given the help they may require to apply for them. Other patients report battling for access to benefits to which they are entitled. This may stem from the relevant authorities lacking awareness of rare diseases and the challenges patients face with these conditions not "fitting easily into boxes".

People affected by rare diseases can suffer financially as a result of, for example, travel costs to and from multiple hospital appointments, the need for expensive equipment or home modifications, having to give up employment or take time off work as a result of their condition, or having to reduce working hours to care for a patient with a rare condition. In RDUK's recent survey 61% of those who cared for a person with a rare disease said that their role as a carer affected their ability to hold paid employment. Financial difficulty adds an extra worry and stress to families and they should be helped to access the social and financial support needed.

### Recommendation 4

**Social support for those affected by rare diseases should be a fundamental part of the patient's care package. Patients and/or their families should be assisted in accessing these services and information about them.**

Recommendations 1a, 1d and 1f would also help to address this problem.

One stop clinics are discussed in Recommendation 3a, Delivering Coordinated Care, and would also be beneficial in enabling patients to discuss their non-medical concerns, including financial queries.

# Delivering Coordinated Care

## Current situation

1. Care for patients with rare diseases is often poorly coordinated and fragmented, and there is frequently a lack of communication between all the professionals involved in the care of the patient.
2. Care for patients with rare diseases is not always patient-centred and does not always fully take into account a patient's individual needs and preferences.
3. Care for patients with rare diseases is often not provided holistically and does not always include consideration of their non-medical needs.

## Recommendations

1. There should be a systematic programme of designation for centres of excellence for rare diseases supported by networks linking into local services throughout the UK.
2. Mechanisms should be put in place to ensure good communication between all healthcare professionals involved in the care of a patient.
3. Care for patients with rare diseases should be patient-centred, taking account of an individual's personal needs.
4. Care for patients with rare diseases should be provided holistically, and should include consideration of the patient's and their family's non-medical needs.

## Lack of coordination and communication in patient care

### Current situation

**Care for patients with rare diseases is often poorly coordinated and fragmented, and there is frequently a lack of communication between all professionals involved in the care of the patient.**

Most rare conditions affect multiple body systems, meaning that many professionals from different specialties and disciplines need to be involved in the care and treatment of the patient. As such, it is vital that there are formal links between all those involved to ensure that the patient is receiving the best possible care and that it is optimally coordinated. Often, however, these links are not in place or communication is weak. This may be because there are restrictions in place that prevent communication between professionals, such as strict confidentiality and access regulations, or it may be that professionals do not know who they should be talking to or this responsibility is not included in their job description, and so specific time is not allocated to this communication.

It is also too frequently the case that patients meet professionals who have not previously been informed of their condition or situation. Patients and families are therefore often obliged to re-tell their story time and again to each new specialist they see. This can be time-consuming, and is not the best use of a patient's or specialist's time, especially if either has had to travel a considerable distance to attend an appointment. The result is an inefficient use of NHS resources, as well as reduced patient satisfaction.

Fragmented care can result in patients feeling 'lost in the system', which leads to a lack of faith in the healthcare system and dissatisfaction with the services they receive. Poor coordination and communication make it difficult for the patient to keep track of when they should be visiting which professional, resulting in missed opportunities for receiving vital care and support. It also makes it difficult for patients to identify who to go to for help with a specific problem or question and where to access the care they need. Often it



has resulted in patients 'rattling around in the system', with each professional they see looking at specific elements of a patient's condition, but no-one being concerned with the condition as a whole.

Fragmented care can also result in added expenditure to the NHS, due to services being used inefficiently and not making the best of professionals' and patients' time. Poorly coordinated care makes forward planning more difficult and can result in missed opportunities for interventions, sometimes leading to costly alternative treatments further down the line or avoidable emergency hospital admissions.

### **Recommendation 1**

**There should be a systematic programme of designation for centres of excellence for rare diseases supported by networks linking into local services throughout the UK.**

The EU criteria for designation as a centre of excellence require the centre to have the following:

- sufficient activity and capacity to provide relevant services at a sustained level of quality
- capacity to provide expert advice, diagnosis or confirmation of diagnosis, to produce and adhere to good practice guidelines and to implement outcome measures and quality control
- multi-disciplinary approach
- high level of expertise and experience, as documented through publications, grants or honorific positions, teaching and training activities, etc
- strong contribution to research
- involvement in epidemiological surveillance, such as registries
- close links and collaboration with other expert national and international centres, and capacity to network
- close links and collaboration with patient associations, where they exist
- appropriate arrangements for patient referrals from other EU countries
- appropriate capacities for diagnosing, following-up and managing patients, with evidence of good outcomes, where applicable.<sup>30</sup>

We believe that centres of excellence for rare diseases should be developed in the UK in accordance with these criteria. These centres need not require the development of new physical buildings, rather organisation and further development of services that already exist. This would result in the development of centres that have expertise in rare conditions and would be able to centrally coordinate care of patients with that disease. They would have in-depth knowledge of the implications of the condition and would know what services are likely to be required and when.

The staff in centres of excellence should consist of experts in the particular condition(s) and would give guidance and advice, based on experience, to patients and families affected by these conditions and the professionals involved in their care. They would be able to provide information on the best care pathways for the group of diseases and to aid communication and coordination of care.

Centres of excellence would also be ideally placed to develop accredited, agreed guidelines of care for the condition(s) that they represent. This would again help in the coordination of a patient's care as there would be an agreed standard from which to work and create care plans for individual patients.

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30. 'Centres of Reference for rare diseases in Europe: State-of-the-art in 2006 and recommendations of the Rare Disease Task Force', Rare Disease Task Force, December 2006



## Care guidelines: an example of good practice

The family guides produced by TREAT-NMD for neuromuscular conditions such as Duchenne Muscular Dystrophy are an excellent example of a care guideline. They are based on the detailed international consensus guidelines produced by a group of 84 neuromuscular disease experts and published in *Lancet Neurology*<sup>1</sup>. These guidelines for best practice are an excellent tool for healthcare professionals, with the family guides in place to inform families of the likely progression of DMD and of the care they should expect and are entitled to.

Dyscerne, a European network of centres of expertise for dysmorphology, have also produced management guidelines for four conditions – Angelman syndrome, Kabuki syndrome, Noonan syndrome and Williams syndrome<sup>2</sup>. The guidelines were produced following a systematic evidence review by a multidisciplinary group of clinicians to identify the optimum management of these conditions. The guidelines have been very well received by patients and professionals, and now that the methodology is in place for their production, we recommend that it be used to produce similar guides for other conditions.

<sup>1</sup> [www.treat-nmd.eu/downloads/file/standardsofcare/dmd/lancet/the\\_diagnosis\\_and\\_management\\_of\\_dmd\\_lancet\\_complete\\_with\\_erratum.pdf](http://www.treat-nmd.eu/downloads/file/standardsofcare/dmd/lancet/the_diagnosis_and_management_of_dmd_lancet_complete_with_erratum.pdf)

<sup>2</sup> <http://www.dyscerne.org/dysc/Guidelines>

The central coordination provided by these centres would improve a patient's care and have a positive impact on their quality of life and wellbeing by ensuring that care was less fragmented.

Having this central coordination would also lead to savings to the health system as care would become more consistent, there would be a reduction in unnecessary appointments and interventions, and it would allow for forward planning to improve health outcomes while reducing the need for costly emergency hospital admissions.

As defined in the EU criteria, centres of excellence would be able to develop links with other expert centres, so allowing the development of networks, which would also benefit collaboration in research into rare diseases. This network development ties in with Recommendation 1a of the Coordination of Research section of this report.

### Specific actions to be taken to achieve the development of rare disease centres of excellence include:

- a. Rare diseases should be grouped based on the needs and clinical features of conditions and centres should be developed for each grouping. We recognise that it would be impossible to develop centres of excellence for each of the 6,000 individual rare diseases, therefore this approach would maximise the resources of such a centre of excellence and prevent duplication of work. The grouping of rare diseases and development of centres of excellence in this manner ties in with Recommendation 4a in the Commissioning and Planning section of this report.

Centres of excellence would then feed information into regional centres dispersed according to geographical and population needs.

As an example of an existing model, the Muscular Dystrophy Campaign (MDC) has supported the development of Muscle Centres in the UK, which has allowed the growth of multidisciplinary clinics alongside specialised research units. With charitable funding, networks between centres have developed to support professions allied to medicine and national audit, as well as patient registries (as suggested in Recommendation 3a of the Coordination of Research section of this report). The centres and network in the UK are supported by an international network and internationally agreed care guidelines. Examples such as this should be used as models of good practice on which to base new centres and networks.

Other possible groupings of conditions could include congenital malformations in children where management issues and considerations for follow up are similar, and rare autoimmune diseases to ensure sharing of novel treatment options.

- b. Funding for rare disease centres of excellence should be provided by a specific budget, which should be directly allocated to the specialised commissioning or planning body (as described in Recommendation 3a

of the Commissioning and Planning section of this report).

There needs to be a mechanism in place to ensure that development of these centres of excellence is based on the clinical need of the UK, rather than on who 'shouts the loudest'. This is further discussed in Recommendation 4b of the Commissioning and Planning section of this report. A system should then be established to roll out these centres, building on centres of expertise that already exist, and relating to the needs base that is identified.

- c. The role of patient organisations in the development and implementation of the work of centres of excellence should be clearly defined. We recommend that, although the patient organisation and the centre of excellence each maintain their own discrete role in order best to achieve their own aims and objectives, there should be some functional integration between the two to optimise the benefits available to the patient. Patient organisations are ideally placed to liaise between newly diagnosed patients and the centre of excellence to ensure that all patients are aware of the centre's existence and that they are accessing the services that the centre is able to provide. There should be a core set of responsibilities agreed between the relevant patient organisations and centres of excellence to optimise this liaison, but there must also be a level of confidentiality maintained within each to ensure that patients feel assured that their information within each is secure.

## Recommendation 2

### **Mechanisms should be put in place to ensure good communication between all healthcare professionals involved in the care of a patient.**

Centres of excellence as discussed in Recommendation 1 would ensure that all professionals involved in the care of a patient are aware of each other's role and input into the patient's care, and could work together to achieve the most effective care plan. Improved communication between all professionals would ensure better coordination of care and would also encourage thought around the care of the patient's condition as a whole, rather than by organ or body system. Increased communication between specialists would ensure that each had a thorough understanding of the patient and their condition, and would enable sharing of useful information between professionals, thereby improving the experience of the patient.

#### **Specific actions that should be taken to improve communication between professionals include:**

- a. A 'hub and spoke' model of communication between centres of excellence and local services should be established. This model of communication would enable expert information from the centre of excellence to be fed out to the professionals delivering care in the patient's local centre, to enhance communication between these professionals and knowledge of each other's roles. This 'hub and spoke' model has previously been described in the Prevention and Diagnosis section of this report (Recommendation 2c) to increase healthcare professionals' knowledge and awareness of rare diseases.
- b. Every patient with a rare, chronic disease should have an agreed, personalised, and regularly updated care plan, which should be agreed between the centre of excellence/specialised service and local services. This care plan should set out drug, therapy and follow up regimens and ideally be linked to standard treatment guidelines for the condition. Implementation of the care plan should be overseen by the Care Coordinator (Recommendation 3b).

Having a care plan in place would stop patients from feeling 'lost in the system' as they would be assured that there is a plan detailing what services and support they should be receiving and when. This would help empower patients to enforce their right to the high quality care that they should be receiving if they do not feel they are receiving it.

Development of a care plan for a patient would encourage communication between professionals, and between the centre of excellence and the patient's local service. This would ensure that everyone involved in the care of the patient was aware of each other's role and how it fitted into the care plan of the patient.

- c. In collaboration with the centres of excellence, regular meetings should be held between the professionals involved in the care of rare disease patients, to discuss their care plan and the best ongoing course of action.

## Multidisciplinary team meetings: an example of good practice

St. Mary's Hospital in Manchester has many multidisciplinary team meetings that often involve members of the genetics team and other specialists. These meetings are used to discuss particular patient cases, and also help to assess whether a patient should be referred to a geneticist for a separate appointment. These meetings have been shown to be an effective use of professionals' time and NHS resources as many cases can be discussed in one meeting, and fewer patients are unnecessarily referred for a genetics appointment.

We recommend that the model of these meetings be rolled out in other hospitals.

- d. 'Flagging systems' should be introduced to provide a link between hospital admissions systems and the patient's lead consultant. A system exists in North Bristol Trust and University Hospitals Bristol Trust in which consultants elect to be informed about the admission of patients with specific conditions, usually those patients with a rare, chronic, life-threatening condition, and they then receive an automated email to inform them if the patient is admitted to hospital. The email alerts the consultant to the fact that their patient is in the hospital, and enables them to make contact with the ward staff in charge if the consultant thinks there is anything they should be aware of in regard to the patient's condition. This model works well and is a cost-effective, simple method to improve communication within hospitals.

We therefore recommend that a similar system be introduced in other UK hospitals.

- e. Systems should be put in place to enable better sharing of information on rare disease patients between all professionals involved in their care. Often access rights to hospital records of patient information can prevent specialists from different disciplines accessing patient notes to identify what care or treatment they are receiving from another department. This therefore restricts communication between professionals and can inhibit them from being aware of aspects of the patient's care that it may be beneficial for them to know about.

We recognise that patients may want part of their care to remain private, therefore we recommend that a system of consent be established that enables a patient to agree to sharing their notes between departments, and allows this access once consent is given. A system such as this, whereby all patients' information can be accessed from any NHS computer, once patient consent has been given, would facilitate communication between all professionals involved in the care of rare disease patients, and would ensure that they were aware of all information that might influence their decision on treatment or therapy options. Furthermore, patients should be able to access this information to enable them to gain a full overview of the care they are receiving, as well as all their test results and other medical information.

- f. Patients should be given the option of being provided with copies of their own medical records. Patient-held records would be kept up-to-date by patients and the professionals they see, and patients could take these records with them to each appointment to ensure that all the specialists they see have access to their most recent medical notes. As well as ensuring that specialists have all the information they need, this would also give some control back to the patient and make sure that they know all the details of their own condition. The use of patient-held records, or an online system of sharing patient notes as discussed in Recommendation 2e would prevent patients and families from constantly having to repeat themselves and would also ensure that, if patient notes were not passed on to a specialist prior to an appointment, the patient could hand over their records complete with all their notes. We therefore recommend that a generic outline of the patient-held record be produced and trialled for use as a pilot project for patients with a particular condition(s) with a view to expanding this.

## Lack of patient-centred care

### Current situation

**Care for patients with rare diseases is not always patient-centred and does not always fully take into account a patient's individual needs and preferences.**

Often, some aspects of care for patients with rare diseases are not, or cannot, be provided locally due to the rarity of the condition, meaning that specialists are located around the country. Patients may therefore need to travel long distances to visit the various specialists associated with different aspects of their condition. While in our experience patients are willing to travel long distances if they are then able to see all the

specialists they need to in one go, travelling around the country for multiple different appointments can be hugely disruptive, and costly, to patients and their families.

RDUK's recent survey of patient and family experiences found that 66% of patients reported having to travel for over an hour to get to their furthest clinic, of which 32% had to travel for over two hours, with 15% having to travel over three hours. 25% of rare disease patients have to attend three or four different clinics for their condition, with a further 12% having to attend more than five different clinics. This obviously has implications for a patient or carer's ability to hold down employment or attend school consistently, with many patients and carers reporting that they had to leave their job, or significantly reduce their working hours, to attend all appointments. Travelling can also be very costly for families, which can further add to the financial burden of the condition. With more consideration of a patient's needs and situation, some patients can be supported to continue to live their lives as normally as possible.

### Recommendation 3

#### **Care for patients with rare diseases should be patient-centred, taking account of an individual's personal needs.**

Patients should be supported to live their lives as normally as possible, and should not have to educate and inform all the professionals with whom they come into contact. Care should be provided in a way that is respectful of an individual's needs and, if possible, should not disrupt their daily life and routine.

#### **Specific actions that should be taken to make care for rare disease patients more patient-centred include:**

- a. Specialist clinics within centres of excellence and, where appropriate, joint clinics of multiple specialists should be developed and supported for rare diseases. These clinics act as a 'one stop shop' where patients can access all the specialists they need to see in one visit. This reduces the need for patients to travel to multiple different clinics, causing less disruption to patients and their families. The clinics also help to ensure coordination and communication between all members of the multidisciplinary team involved in the patient's care whilst making the most effective use of consultants' time. Patients have told us that they are willing to travel further to clinics if they are able to see all the people they need to and receive high quality care, thereby reducing the need to travel to multiple different appointments.

Existing specialist clinics, such as those commissioned by the National Specialised Commissioning Team in England have been proven to be successful and beneficial to the patients who attend them. We recommend that the model of these clinics be replicated for other conditions. The benefits that these clinics can provide to a patient's care, well-being and satisfaction, as well as the increased efficiency provided by such a clinic, should be considered as a balance to offset any initial investment that development may incur. Clinics such as this have previously proven to be cost-neutral or even cost-saving.

Specialist clinics should be developed as a part of each relevant centre of excellence so that the experts from the centre, in conjunction with input from the relevant patient organisation could identify which healthcare professionals would be needed at the clinics and what needs of the patients should be addressed. Patient organisations may be able to provide psychological support, social support and information on relevant benefits for which the patient may be eligible.

Joint clinics of professionals within a hospital may also sometimes provide many benefits for patients by acting in a similar way as specialist clinics.

#### **Joint clinics: an example of good practice**

Guy's Hospital, London, runs combined clinics for 12 different rare genetic conditions. The format of each clinic varies, but includes attendance by the genetics team and multiple specialists involved in the condition. The clinic is usually coordinated by a genetic counsellor, and following attendance the patient receives a letter summarising the outcomes of the clinic and the results of any tests. Patients are given an emergency contact number to use at any time between clinics.

This is a successful model, and funding should be provided for this type of service to ensure its sustainability. Participation in these clinics should be included in the health professional's job description and specific time should be allocated to carrying out this role.

Joint clinics and specialist clinics have other benefits too, especially for training and for research, but we would like to avoid being prescriptive about how they are organised. Such clinics can be tailored creatively to meet particular local needs and take advantage of local facilities. Coordinated appointments arranged in the same hospital on the same day may also achieve many of the goals of these clinics.

### Specialist clinics: examples of good practice

In April 2010 the Laurence-Moon-Bardet-Biedl Society received funding for nationally commissioned specialist Laurence-Moon-Bardet-Biedl syndrome (LMBBS) clinics. Twelve of these clinics a year are now being held per year, in London or Birmingham. Patients are initially invited by the LMBBS Society, followed by an invitation from the relevant hospital. On the day of the clinic the Society provides patient information packs and Society Support Workers are also in attendance to help patients. Patients attend either in the morning or afternoon, during which time they see all relevant specialists including the genetics team, an ophthalmologist, a urologist, a psychologist, an endocrinologist and a dietician. Throughout the session the patient stays in the same room and the specialists rotate to them. Following the clinic, clinicians' reports are collated by the hospital and copies are sent to the patient, their GP and their local hospital. Overnight accommodation and travel to the clinics can be arranged for patients, and it is hoped that this will become an annual visit for them. Patients from all four home nations are invited to attend, to be funded by the relevant specialised services bodies.

These clinics have already been very successful, with patients benefiting from this arrangement and providing positive feedback, such as the following comments:

- 'The clinic and the whole set-up of the day was excellent... We really feel that we could not have received better care.'
- 'Great to see so many specialists on the one visit and everyone was very understanding and patient... All day we were treated very well and never rushed and had lots of time to ask questions.'

Similar nationally commissioned clinics exist for Alstrom syndrome, in Torbay Hospital for adults and Birmingham Children's Hospital for children. Patients attending these clinics have access to multiple medical tests and assessments with relevant specialists in the morning, and the afternoon is designated to discussing non-medical issues including social, educational and psychological needs. Patients value this structured review and the opportunity to see all doctors in one hospital visit, as well as having time dedicated to discussing all other aspects of the condition and to meet with other patients.

These are just two examples of where this model of clinic has been successful and gratefully received by patients and their families. We recommend that these models be rolled out for other relevant rare diseases to enable more patients to have the same experience.

- b. Patients with rare diseases should be offered a designated Care Coordinator to liaise between themselves and the services they use, and to ensure that the right services are brought together at the right time. The Care Coordinator should be supported by the relevant centre of excellence to provide them with expert medical advice and the necessary knowledge of the condition. The Care Coordinator should assist in liaising between the different professionals involved in the care of a patient, again making sure that services are used more effectively.

Improved linkage between centres of excellence and local services (as discussed in Recommendation 2a) would enable communication between the centre of excellence and the Care Coordinator to advise them on the best care pathways, so that more of the patient's care can be delivered in the local area and travelling time is kept to a minimum. This linkage would enable more care to be provided closer to the patient's home, leading to more patient-centred care and the most efficient use of NHS resources. As well as benefiting the patient, this information sharing would also raise professionals' awareness of particular diseases, assisting them in the identification and management of any new cases.

Care Coordinators already exist for other types of conditions, including mental health conditions. Here, the role may be taken by a social worker, occupational therapist or a community mental health



nurse. Recently published NICE guidelines for cancer of unknown primary (CUP) origin<sup>31</sup> recommended that 'every hospital with a cancer centre or unit should assign a CUP specialist nurse or key worker to patients'. The role of this person would be to coordinate patients' care, to liaise with GPs and support services, to ensure that patients and carers have access to information, advice and support, and to be in regular contact with the patient and be their advocate when necessary. If roles such as this are possible for these conditions, we believe that they should also be made available to the rare disease patients who need them. Patients with rare diseases are often in particular need of Care Coordinators due to the often multi-system nature of their conditions and poor levels of knowledge and awareness of how best to manage them.

The Care Coordinator should be a trained professional whose role is to ensure that a care plan is in place and acted upon. The Care Coordinator's role should include being available to talk to the patient about his or her concerns and giving consideration to the needs of the family or carer. The person best equipped to carry out the role of Care Coordinator would vary between conditions but suggestions of appropriate professionals have included specialist nurses working within the appropriate fields, members of the genetics team or social workers. Where the role of Care Coordinator is additional to a professional's existing role, it should be included in their job description and specific time should be allocated to carrying out this role.

Care Coordinators should be able to make appointments for patients and schedule multiple appointments for the same day. There should be a system that gives priority to booking these coordinated appointments, thereby reducing the need for a patient to travel to hospital on multiple occasions.

### **Patient Organisations' role in supporting Care Coordinators: examples of good practice**

Patient organisations have played an important role in the provision of this service, though to date none can provide a comprehensive universal service due to a lack of resources. As an example, the Tuberous Sclerosis Association Specialist Advisers provide support, advice and information to patients over the telephone, via email or during home visits. They also liaise with professionals involved in the patient's care to ensure the best treatment, and address the issues of families and carers. The Progressive Supranuclear Palsy (PSP) Association employs Development Officers in all four home nations to support patients and families, and to educate and encourage local service providers to give excellent care to PSP patients. The MDC Care Advisors carry out a similar role for neuromuscular disease patients (see Recommendation 1f in the Patient Care, Information and Support section of this report).

Where this service is being provided by patient organisations, we recommend that funding should be provided by the UK health departments to support this core service, with adequate coverage based on the understood prevalence of the conditions. The MDC was recently awarded 50% funding from the NHS for their Care Advisors, to be fully funded by April 2011. This model should be built on for other conditions where there is not already someone in post within the healthcare system to fulfil this role.

Recommendations 2e and 2f would also contribute to more patient-centred care by ensuring that when a patient attended an appointment, the healthcare professional was aware of their situation and the patient would not have to spend time explaining their whole medical history.

31. 'Diagnosis and management of metastatic malignant disease of unknown primary origin', Full guideline, Developed for NICE by the National Collaborating Centre for Cancer, July 2010



## Lack of holistic care and consideration of non-medical needs

### Current situation

#### Care for patients with rare diseases is often not provided holistically and does not always include consideration of their non-medical needs.

The Patient Care, Information and Support section of this report outlines the problems experienced by patients in terms of lack of support with their non-medical needs.

Support for the needs of families and carers of patients with rare diseases is also often lacking and must be addressed. Caring for a loved one can be a very difficult job, and so adequate support must be provided for families and carers.

A prime example of the lack of holistic care is seen in the transition from paediatric to adult services. Patients with rare diseases often experience problems with medical, psychological, financial and social issues at this time, and too often support in these areas is not available for them to access.

Problems at service transition include the fact that patients often feel unsupported when they enter adult services as they have to leave the care of a paediatrician with whom they may have developed a strong bond and there is no equivalent person to take overall responsibility for the patient in adult services. In leaving children's services they may lose out on access to children's centres where they had been able to access medics and therapists at one time, contact with therapists at school, some aspects of primary care in effect being provided by the paediatrician, as well as the overall coordination of care by the paediatrician. There is often little or nothing equivalent to compensate for the lack of these services for adults.

We have been informed that from the perspective of a patient with a rare disease, 'when you are in paediatric services, you are treated as a whole person; when you go to adult services, you are just a series of organs and body systems'<sup>32</sup>.

Many patients are not given a discharge pack or any information to pass to adult services, and as such they may find that the new health professionals with whom they are dealing lack knowledge of their particular rare condition.

However, it is not just problems with medical care that patients and families or carers often experience at transition. Patients and families have reported feelings of isolation and being 'cut off' following transition and then either not being offered, or having to fight for, necessary psychological support. We have also been informed of patients and families no longer being able to claim benefits following transition to adult services and not being offered any support or information in this regard.

### Recommendation 4

#### Care for patients with rare diseases should be provided holistically, and should include consideration of the patient's and their family's non-medical needs.

Patients' psychological, social, financial and any other needs should be addressed as well as their medical needs. Patients or their families should not have to fight for this support. It should be included as an integral part of their care plan and services should be readily accessible if and when they are needed.

Care must be taken to ensure that structures that were in place to care for and support a patient under paediatrics continue to be made available post transition. Care must not stop, or reduce in quality, when a patient enters adult services, and the clinicians they encounter following transition must be fully aware of that patient's situation. Care must be taken to ensure that the process of transition is as straightforward as possible and that the care received in adult services takes into account the patient as a whole, rather than just each aspect of the condition. Resolving issues surrounding transition from paediatric to adult services is especially important because, due to better care and understanding of the condition, an increasing number of children with rare diseases are surviving into adulthood.

#### Specific actions to be taken to ensure that care is more holistic include:

- a. The delivery of appropriate psychological, social, financial and any other care should be included

32. Quote from patient representative during RDUK Working Group meeting.

within the care plan produced by the centre of excellence and the patient's local service. The centre of excellence would be aware of the common non-medical needs of patients affected by the particular condition(s) in which they specialise, and should liaise with the patient's Care Coordinator to ensure that these needs are considered thoroughly. Delivery of this support should feature as an integral aspect of a patient's care.

- b. Best practice care guidelines produced by the centres of excellence (as suggested in Recommendation 1), in collaboration with others such as patient organisations, should include how to coordinate transition from paediatric to adult services. The guidelines should describe how best to deal with issues that may arise, and what services are likely to be required following transition. These guidelines would enable a proactive approach to care, and help to ease transition by highlighting areas that may cause concern so that they can be addressed in good time.
- c. The needs of carers and families should also be considered and addressed. This too should be incorporated into the patient's care plan and support should be made available if and when it is needed. This may include respite care for carers and families to help them feel better equipped to care as best they can.

The recommendations for a Care Coordinator (Recommendation 3b) and for patient-held records (Recommendation 2f) would also help to ease transition for patients. The information prescription, as described in Recommendation 1a of the Patient Care, Information and Support section of this report should also help to enable a more holistic approach to care.



# Table of Abbreviations

- AGNSS – Advisory Group for National Specialised Services
- AMRC- Association of Medical Research Charities
- Array-CGH – array comparative genomic hybridisation
- ASBAH – Association of Spina Bifida and Hydrocephalus
- BME – Black and minority ethnic
- BPSU – British Paediatric Surveillance Unit
- BSHG – British Society for Human Genetics
- CBD – Cortico Basal Degeneration
- Climb – Children Living with Inherited Metabolic Diseases
- CPD/CME – Continuing Professional Development/ Continuing Medical Education
- CRN – Clinical Research Network
- CUP – Cancer of unknown primary
- DDD – Deciphering Developmental Disorders
- DH – Department of Health
- DLA – Disability Living Allowance
- EHDN – European Huntington’s Disease Network
- EMA – European Medicines Agency
- EME – Efficacy and Medical Evaluation
- GP – General Practitioner
- HD – Huntington’s Disease
- HSCB – Health and Social Care Board
- HTA – Health Technology Appraisal
- ICD – International Classification of Diseases
- IFR – Individual Funding Request
- IRAS – Integrated Research Application System
- LMBBS – Laurence-Moon-Bardet-Biedl syndrome
- MCADD – Medium Chain acyl-CoA Dehydrogenase Deficiency
- MDC – Muscular Dystrophy Campaign
- MHRA - Medicines and Healthcare products Regulatory Agency
- MRC – Medical Research Council
- NGS – Next Generation Sequencing
- NHS – National Health Service
- NICE – National Institute for Health and Clinical Excellence
- NIGB – National Information Governance Board
- NIHR – National Institute of Health Research
- NRES – National Research Ethics Service

- NSC – National Screening Committee
- NSD – National Services Division
- PALS – Patient Advice and Liaison Service
- PCD – Primary Ciliary Dyskinesia
- PCT – Primary Care Trust
- PDCO – Paediatric Committee
- PIP- Paediatric Investigation Plan
- PSP – Progressive Supranuclear Palsy
- QALY – Quality-Adjusted Life Year
- R&D – Research and Development
- RAPID – Reliable Accurate Prenatal non-Invasive Diagnosis
- RCF – Rarer Cancers Foundation
- RCT – Randomised Controlled Trial
- REC – Research Ethics Committee
- SCG – Specialised Commissioning Group
- SMC – Scottish Medicines Consortium
- SSNDS – Specialised Services National Definitions Set
- UKGTN – UK Genetic Testing Network
- VBP – Value Based Pricing

# Annex 1: Working Group Members

## Coordination of Research

### Chair – Dr Hilary Burton, PHG Foundation

- Dr John Crolla, Wessex Regional Genetics Laboratory
- Professor Dian Donnai, University of Manchester, Manchester Academic Health Sciences Centre
- Professor Tony Holland, Cambridge Intellectual and Development Disabilities Research Group
- Richard Lynn, British Paediatric Surveillance Unit, Royal College of Paediatrics and Child Health
- Aruni Mulgirigama, Pfizer/Orphan Diseases Industry Group
- Becky Purvis, Association of Medical Research Charities
- Dr Eamonn Sheridan, University of Leeds
- Dr Mark Taylor, Birmingham Children’s Hospital
- Dr Adrian Thrasher, UCL Institute of Child Health
- Susan Walsh, CGD Research Trust
- Professor David Wield, ESRC Innogen Centre, University of Edinburgh

## Prevention and Diagnosis

### Chair – Dr Jim Bonham, Sheffield Children’s NHS Foundation Trust

- Judy Birch, Pelvic Pain Support Network
- Angela Burgess, Wales Gene Park
- Professor Jill Clayton-Smith, University of Manchester
- Jane Fisher, Antenatal Results and Choices
- Dr Zosia Miedzybrodzka, University of Aberdeen
- Professor Chris O’Callaghan, Leicester Royal Infirmary
- Steve Potter, Genzyme Therapeutics
- Dr Nadeem Qureshi, University of Nottingham
- Pat Roberts, Save Babies Through Screening Foundation UK
- Professor Peter Soothill, Bristol University
- Dr Su Stenhouse, UK Genetic Testing Network/ Glasgow Yorkhill Hospital

## Patient Care, Information and Support

### Co-Chair – Lesley Greene, Vice-President, Climb and Eurordis Patient Rep

### Co-Chair – Andrew Wilson-Webb, Rarer Cancers Foundation

- Tariq Ahmed, Bradnet
- Sandy Craine, CML Support
- John Dart, DEBRA
- Louise Derbyshire, Contact a Family
- Brian Ellis, Heart UK
- Lyn Inman, Muscular Dystrophy Campaign
- John Irwin, Actelion/Orphan Diseases Industry Group
- Professor Maggie Kirk, University of Glamorgan
- Christine Lavery, MPS Society
- Prisca Middlemiss, Unique
- Kathy Oliver, International Brain Tumour Alliance
- Bill Owen, Niemann Pick Disease Group
- Dr Christine Patch, Guy’s and St Thomas’ Hospital
- Beverly Searle, Unique
- Margaret Smith, University of Dundee School of Nursing and Midwifery
- Sophie Thomas, MPS Society



## Delivering Coordinated Care

### Chair – Dr Atul Mehta, Royal Free Hospital, London

- Jon Beauchamp, Alexion/Orphan Diseases Industry Group
- Professor Kate Bushby, University of Newcastle
- Professor Angus Clarke, Cardiff University
- Dr Peter Galloway, Royal Hospital for Sick Children, Glasgow
- Pam Griffiths, University of Manchester
- Rachel Lewis, Royal Free Hospital, London
- Rea Mattocks, Uveitis Information Group
- Marie McGill, Single Gene Complex Needs Project
- Kay Parkinson, Alstrom Syndrome UK
- Daniela Pilz, University Hospital Wales
- Linda Richfield, Royal Free Hospital, London
- Heather Skirton, University of Plymouth
- Sir Bert Massie CBE, The Compact (Guest attendee)

## Commissioning and Planning

### Chair – Adrian Pollitt, former Director of National Specialised Commissioning

- Steve Collins – National Commissioning Group
- Dr Peter Corry, Bradford Hospitals
- Jane Deller, UK Genetic Testing Network
- Josie Godfrey, National Specialised Commissioning Team (replaced Steve Collins)
- Dr Tom Kenny, South Central Specialised Commissioning Group
- Eric Low, Myeloma UK
- Dr Jane Lucas, Southampton General Hospital
- John Murray, Specialised Healthcare Alliance
- Chris Pickard, Pfizer/Orphan Diseases Industry Group
- Amy Pott, Baxter/Orphan Diseases Industry Group
- Helen Rainbow, Macmillan Cancer Research
- Sophie Wintrich, MPS UK Patient Support Group (Guest attendee)
- Rodney Taylor, MPS UK Patient Support Group (Guest attendee)

# Annex 2: Respondents to RDUK Consultation

## Patient Organisations

- ACT
- Advocacy for Neuroanthocytosis Patients
- AKU Society
- Amy and Friends
- Association for Spina Bifida and Hydrocephalus
- Association of Medical Research Charities
- AT Society
- Behcet's Syndrome Society
- Breathtakers
- chILD Lung Foundation UK
- Chromosome 18 Registry and Research Society
- Climb
- Help the Hospices
- Hypermobility Syndrome Association
- Jennifer Trust for SMA
- Lymphangiomatosis and Gorham's Disease Alliance
- Motor Neurone Disease Association
- Muscular Dystrophy Campaign
- Myasthenia Gravis Association
- Myrovlytis Trust
- Niemann Pick Disease Group UK
- Picks Disease Association
- Post Polio Network
- Prader-Willi Syndrome Association UK
- PSP Association
- Raynaud's and Scleroderma Association
- Relapsing Polychondritis Support Group
- The British Thoracic Society
- The CGD Research Trust
- The Fragile X Society
- The Ring Chromosome 20 Foundation
- The Society for Mucopolysaccharide Diseases
- Vasculitis UK
- Waldenstrom's Macroglobulinaemia UK

## Pharmaceutical/Biotech Companies

- Actelion
- Bayer
- BioMarin
- Bio Products Laboratory
- Genzyme Therapeutics
- Orphan Disease Industry Group (Umbrella body)
- Shire HGT

## Healthcare Professionals

- Kate Bushby, TREAT-NMD
- Angus Clarke, Consultant in Clinical Genetics, St David's Hospital, Cardiff
- Dr Shelagh Joss, Consultant Clinical Geneticist, Royal Hospital for Sick Children, Glasgow
- Carolyn Owen, Genetic Counsellor, Wrexham Maelor Hospital
- Gill Rumsby, UCL Hospital
- Dr Bernd Schwahn, Consultant in Paediatric Metabolic Medicine, Royal Hospital for Sick Children, Glasgow
- Dr Claire Shovlin, Consultant, Imperial College Healthcare NHS Trust
- Stuart Tanner, Professor of Paediatrics, University of Sheffield
- Dr Mark Taylor, Consultant Paediatric Nephrologist, Birmingham Children's Hospital
- Patricia Woo, Professor of Paediatric Rheumatology, on behalf of MCRN/Arthritis Research UK Paediatric Rheumatology Clinical Studies Group
- Genetic Counsellors of Guy's and St Thomas' Hospital, London

## Researchers/Academics

- Jane Cox, Deputy Head of Research, Sussex Kidney Unit, Royal Sussex County Hospital
- Ruth Charlton, Scientific Director of Molecular Genetics, Yorkshire Clinical Genetics Centre
- Sophie Duport, Head of Research, The Royal Hospital for Neurodisability
- Kat Hill, Clinical Genetics Research Assistant, Clinical Genetics Dept., Royal Devon and Exeter Hospital
- Alison Kerr, Rett Syndrome Register
- Dr Anil Mehta, University of Dundee
- Jane Salotti, Research Associate, Institute of Health and Society
- David Wield, ESRC Innogen Centre, University of Edinburgh
- Professor Sir Ian Wilmot, Director, MRC Centre for Regenerative Medicine

## Professional Bodies/Networks

- Association of Genetic Nurses and Counsellors
- British Society for Allergy and Clinical Immunology
- Clinical Genetics Society
- European Huntington's Disease Network
- International Network of Paediatric Surveillance Units
- Joint Committee for Medical Genetics (The Royal College of Physicians, The British Society of Human Genetics, The Royal College of Pathologists)
- Society for Endocrinology
- The Royal College of Physicians
- UK Primary Immunodeficiency Network
- UK Working Party for Rare and Orphan Lung Disease

## Public Bodies

- UK Genetic Testing Network
- UK Newborn Screening Committee

## Patient/Relative

- Jane Alcock, parent of a rare disease patient
- Anna Allford, patient
- John Booth, parent of a rare disease patient
- Kate Brindley, parent of a rare disease patient
- Jim Brown, husband of rare disease patient
- Wendy Davey, parent of a rare disease patient
- Simon Dendrick, patient
- Lesley Greene, parent
- Kathryn Hennessy, parent of a rare disease patient
- Helena Koval, patient
- Tess Luetchford, patient
- Carol McCulloch, patient
- Gillian Thomas, wife of a rare disease patient
- Maxine Thorne, patient/carrier
- Susan Underwood, patient
- Suzanne Wedlake, parent of a rare disease patient
- Kelly Willoughby, parent of a rare disease patient

## Other

- Specialised Healthcare Alliance (Coalition of organisations and industry)
- Janice Fawell (Individual)
- Jason Maude, Isabel Healthcare (Private diagnosis company)

# Annex 3: Attendees at the RDUK Focus Groups in Devolved Nations

## Wales

- Dr Geoffrey Carroll, Welsh Health Specialised Services Team
- Hayley Cleaver, Chair, Turner Syndrome Support Society
- Professor Steve Dunnett, Cardiff University
- Joe Ferris, ABPI Wales
- Dr Stephen Jolles University Hospital of Wales Cardiff & Vale Health Board
- Professor Marcus Longley, Welsh Institute of Health & Social Care, University of Glamorgan
- Dr Annie Procter, All Wales Medical Genetics Service
- Dr Chris Riley, Strategy Unit, Welsh Assembly Government

## Scotland

- Andrew Deans, Edinburgh Royal Infirmary
- Catriona Johnson, National Services Division
- Dawn Kofie, Scottish Government, Long Term Conditions Unit
- Jane Cox, Genzyme Therapeutics
- Carol Gardiner, Department of Clinical Genetics, Yorkhill Hospital, Glasgow
- Gillian Scott, Department of Clinical Genetics, Yorkhill Hospital, Glasgow
- Marie McGill, NHS Scotland, Single Gene Complex Need Project

## Northern Ireland

- Richard Dixon, Patient and Client Council
- Errol Walsh, Huntington's Disease Association Northern Ireland
- Kathryn Hennessey, Primary Immunodeficiency Association
- Daniel Kelly, Department of Health, Social Services and Public Safety
- Dr Colin Harper, Disability Action
- Professor Helen Dolk, University of Ulster
- Dr Jackie McCall, Public Health Agency
- Aoife Bradley, Department of Medical Genetics, Belfast City Hospital
- Mark Cunningham, Genzyme Therapeutics
- Alison Whann, Genetic Counsellor and Support Worker for the PIA and the Society for Mucopolysaccharide Diseases
- Joanne McOsker, Royal Victoria Hospital
- Dr Pauline Hunter, Muscular Dystrophy Campaign
- Dr Colin Willoughby, Centre for Vascular Science, Queen's University

- Sinead Owens, Disability Action
- Sandra Campbell, PSP Association
- Dr Fiona Stewart, Department of Medical Genetics, Belfast City Hospital
- Terry Hoey, Cavan Tommy Hoey Trust

## Annex 4: Attendees at the EUROPLAN Conference

- Godfrey Adams, Action Duchenne
- Julie Ann Bridge, Pfizer
- Dan Beety, Bayer Health Care
- Dr Tymandra Blewett-Silcock, POPS (Parents of Partially Sighted and Blind Youngsters)
- Valentina Bottarelli, Eurordis
- Professor Kate Bushby, University of Newcastle
- Nick Catlin, Action Duchenne
- Michael Close, Sigma Tau
- Fiona Copeland, PCD Support Group
- Peter Corry, Bradford Teaching Hospitals
- Avril Daly, Fighting Blindness (Ireland)
- William Davis, A-T Society
- Professor Dian Donnai, Nowgen
- Sara Elgott, Orphan Europe
- Derek Elston, ITP Support Association
- Jennifer Freeman, Cavenoma Alliance UK
- Carlee Gilbert, ChILD (Lung) Foundation UK
- Josie Godfrey, National Specialised Services
- Idoia Gomez-Paramio, Nowgen
- Andrew Greaves, Shire Human Genetic Therapies
- Debbie Green, Amy and Friends Cockayne Syndrome UK
- Lesley Greene, Climb
- Alan Heywood-Jones, Alfa Europe Federation
- Dawn Heywood-Jones, Alpha 1 Awareness UK
- Jayne Hughes, Amy and Friends Cockayne Syndrome UK
- Sara Hunt, ALD Life
- Dr Mohit Jain, Talecris
- Dr Ed Jessop, National Specialised Commissioning Team
- Dr Daniel Kelly, Department of Health, Social Services and Public Safety (Northern Ireland)
- Dawn Kofie, Scottish Government (Long Term Conditions)
- Lugdivine Le Dez, Alexion
- Marion McAllister, Nowgen



- Hugh McCarthy, National Rare Renal Disease Registry
- Darren Mansell, Thalidomide Trust
- Anne Mawdsley, Raynaud's Scleroderma Association
- Louise Medus-Mansell, Thalidomide Society
- Dr Zosia Miedzybrodzka, University of Aberdeen
- Janet Mills, Save Babies Through Screening Foundation UK
- Anna Moran, Fighting Blindness Ireland
- Helen Munroe, Advocate Public Affairs
- Paul Mylchreest, Alexion
- Alasdair Nimmo, Myasthenia Gravis Association
- Bill Owen, Niemann Pick Association
- John Parkinson, Alstrom Syndrome UK
- Kay Parkinson, Alstrom Syndrome UK
- Steve Potter, Genzyme Therapeutics Ltd
- Katie Rigg, Multiple System Atrophy Trust
- Dave Roberts, Niemann Pick Association
- Chris Rowland, Pfizer UK
- Graciela Saint, Orphanet
- Dr Beverly Searle, UNIQUE
- Ross Selby, Takeda Ltd UK
- Amy Simpson, Glamorgan University
- Dr Kamlesh Sheth, Swedish Orphan Biovitrum Ltd
- Brian Smith, Glasgow
- Dr John Solly, Myrovlytis Trust
- Dr Shereen Tadros, Great Ormond Street Hospital
- Dr C. Mark Taylor, Birmingham Children's Hospital
- Alan Thomas, Ataxia South Wales
- Gillian Thomas, Wife of husband with rare disease
- Oliver Timmis, Alkaptonuria Society
- Susan Walsh, CGD Research Trust
- Sally Watts, Guy's Hospital
- Richard West, Behcets Society
- Professor David Wild, ESRC Innogen Centre

# Annex 5: Attendees at the RDUK/AMRC Workshop on Rare Diseases

- Professor Robin Ali, University College London
- Julia Ambler, Muscular Dystrophy Campaign
- Mark Bacon, Spinal Research
- Anna Baranski, Specialised Healthcare Alliance
- Ann Marie Barnard, UKCNRC
- Dr Jon Beauchamp, Alexion Pharmaceuticals
- Paul Bingham, Vasculitis UK
- Mike Birtwistle, Health Mandate
- Catherine Bouvier, Net Patient Foundation
- Margaret Bowler, Myotonic Dystrophy Support Group
- Julie Buckler, Action Medical Research
- Anne Carter, Tuberos Sclerosis Association
- Mary-Louise Chiew, Jeffrey Modell FoundationCentre
- Vicki Colledge, Myrovlytis Trust
- Wendy Cook, Nephrotic Syndrome Trust
- Jane Cox, TSA
- John Dart, DEBRA
- Elaine Davies, Kidney Research UK
- William Davis, Ataxia Telangiectasia Society
- David French, Niemann-Pick Disease Group (UK)
- Andrew Greaves, Shire Human Genetic Therapies
- Julie Greenfield, Ataxia UK
- Mike Hales, Alstrom Syndrome UK
- Tess Harris, Polycystic Kidney Disease Charity
- David Hartley, The XLP Research Trust
- Melissa Hillier, Genetic Alliance UK
- Jayne Hughes, Amy and Friends (Cockayne Syndrome Support)
- Carole Ivey, Picks Disease Association
- Annwen Jones, Target Ovarian Cancer
- Stephen Jones, RP Fighting Blindness
- Brian Lovatt, Vision on Rare Diseases
- Daren Mansell, Thalidomide Trust
- Clare McGowne, CSL Behring UK
- Louise Medus-Mansell, Thalidomide Trust

- Dr Anil Mehta, University of Dundee
- John Mills, Vasculitis UK
- Aruni Mulgirigama, Pfizer
- Chris Ogden, ALD Life
- William Owen, Niemann-Pick Disease Group (UK)
- Kirti Patel, Sparks
- Arthur Pearce, Amy and Friends
- Alan Summerside, Thalidomide Trust
- Tom Sunderland, Boehringer Ingelheim
- Sanjay Thakrar, The Myrovlytis Trust
- Lynne Thompson, Breathtakers Charity
- Susan Walsh, Chronic Granulomatous Disorder Research Trust
- Nick Willcox, Myasthenia Gravis Association

## Annex 6: RDUK Management Committee

- Alastair Kent OBE, Director, Genetic Alliance UK (RDUK Chair)
- Steve Potter, Genzyme Therapeutics (Representative of the Orphan Disease Industry Group Partnership) (RDUK Treasurer)
- Becky Purvis, Policy and Public Affairs Manager, Association of Medical Research Charities (RDUK Secretary)
- Dr Peter Corry, Paediatrician, Bradford Hospitals
- Mark Barrett, Alexion (Chair of Orphan Disease Industry Group)
- Dr Marita Pohlschmidt, Head of Research, Muscular Dystrophy Campaign
- Laura Gilbert, Freelance Research Consultant
- Dr Stephen Jolles, Clinical Immunologist, University of Wales Hospital Cardiff (Advisor for Wales)
- Marie McGill, National Lead, Single Gene Complex Need Project (Advisor for Scotland)
- Fiona Stewart, Consultant in Medical Genetics, Belfast City Hospital (Advisor for Northern Ireland)





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