RARE DISEASE | UK

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

Report of Activity

1st July 2011 to 31st December 2012

www.raredisease.org.uk

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A message from the Chair

2012 has seen steady progress towards the production of the National Plan for Rare Diseases for services and support for patients and their families. The consultation, launched on Rare Disease Day 2012, was in itself a first in that it required all the nations of the UK to work together on an issue of common concern in the field of health care. This in itself is recognition that at least some of the needs of patients and families living with rare diseases cannot be met within the confines of any one health care system, but that a broader vision is necessary.

Over 350 consultation responses were submitted to Government. Individuals, institutions, clinicians, patient organisations, industry and many other interested parties took the opportunity to make their views known, and in the process demonstrated a remarkable degree of uniformity in the conclusions they reached. This is hugely important. The number of responses and the consensus around the issues gives a very clear message to Governments across the UK that this is an issue that must be addressed and which cannot be kicked into the long grass and left to moulder. At the start of 2013, we await the launch of the first ever National Plan for Rare Diseases in the UK. Hopefully this will happen sooner rather than later, so there will be the opportunity to look at its implementation before the National Plan is brought back to the Council of Health Ministers of the European Union at the end of the year.

Having played a part in stimulating Governments to act in response to the adoption of the EU Recommendation by the UK Government, we did not sit back and rest on our laurels. RDUK has been constantly active in making the case for change and in generating the evidence to support this. During the year we commissioned a number of reports that will be published in 2013 - on research, care coordinators and the situation of patients and families in Wales and Scotland - and undertook work on a range of issues including access to medicines in Scotland, stimulating debates at Westminster and in the Welsh Assembly and many other things designed to keep the pressure on politicians and policy makers and help them to ensure that the National Plan, when it is published in 2013, is as comprehensive as it needs to be if the NHS is to be able to respond effectively to the legitimate needs of rare disease patients and their families.

RDUK is a coalition of the willing from across the spectrum of stakeholder interests. Membership is open to anyone who shares our goal, and we have experienced steady growth in our numbers during the course of the year from all sectors of the rare disease community. Our members are actively engaged in promoting the aims of this campaign, and we are grateful to them all for their enthusiasm and support. We are also grateful to the staff team supporting the RDUK campaign. Lauren Limb, Stephen Nutt, Samantha Reeve and Alev Sen have played a significant part in securing the progress we have made to date. All have now moved on to new roles elsewhere. We wish them all well, and are grateful for their hard work and dedication to RDUK in the time they have been with us. We also thank and express our appreciation to the members of the management committee who also give their time and expertise to help us shape our direction and our work.

Looking ahead to 2013, we see the opportunity for real progress. We will try hard to realise this, and look forward to working with our members and supporters to bring about not just the publication of the National Plan for Rare Diseases, but also its implementation so the lives of those affected by these conditions can be improved to the fullest extent possible.

Alastair Kent OBE

Executive summary

The 18 months reported here shows we have worked hard to make progress towards a National Plan for Rare Diseases in the UK. We are now an alliance of 1,300 members from all groups of the rare disease community, and over 230 of these are patient groups.

Significant effort in this reporting period has been devoted to ensuring the consultation for the National Plan was published, and that a strong response was made by as many as possible within the rare disease community. We worked across the UK in supporting our members to enable them to respond on the issues that are important to them. We did this through a series of events, information briefings and webinars, which also informed our own response to the consultation. In total, over 350 responses to the consultation were received, a very respectable number.

We have also been active in the home nations building relationships and connections where we can work in partnership to make a difference. In Scotland, a highlight of our activity has been on work around access to medicines; in 2011 we submitted a public petition on access to medicines for orphan diseases to the Public Petitions Committee of the Scottish Parliament. In Wales, we raised awareness of rare diseases at the National Assembly through a short debate on rare diseases. In Northern Ireland, we continue to work closely with NI-based organisations, and are proud of the establishment of the Northern Ireland Rare Disease Partnership.

Other highlights of the year include our work in raising awareness amongst the research community. We will be pursuing these networks to ensure progress is made so that we can build on these to deliver benefits through knowledge and potential treatments that can make a difference.

The year ahead for 2013 looks set to be the busiest yet. We look forward to the publication of the National Plan, where we will be looking for outcomes that can be monitored and measured, and we will be working hard to make sure they are implemented.

'Thank you for your help and thanks for trying to make a difference for us.'
– Kay Anderson, Alpha One Advocacy and Action.

Progress towards the National Plan for Rare Diseases

By July 2011, we had made substantial progress on our campaign. Our two publications – 'Improving Lives, Optimising Resources: A Vision for a Rare Disease Strategy' and 'Experiences of Rare Diseases: An Insight from Patients and Families' - have enabled us to share and discuss our vision for a National Plan for Rare Diseases with various people, including politicians, government officials and decision makers within the NHS. These publications, with their comprehensive recommendations and detailed picture of what life is like with a rare disease in the UK, have provided a sound base upon which to target our efforts to discuss and convince others of why rare diseases matter. As a result, the past 18 months have seen an increase in support and understanding around issues for rare diseases.

It's excellent. The best response I've read.'
– Dr Nick Sireau - Chair of the AKU Society, commenting on RDUK's response to the consultation on the UK Rare Disease Plan.

A large effort for the latter part of 2011 was working alongside our members and all stakeholders across the rare disease community to push for the publication of a consultation on the National Plan for Rare Diseases. We worked tirelessly to raise awareness of the need for a consultation so that that everyone with an interest and an opinion on the area could have their voice heard on the matters that are important to them. We have been in regular contact - working closely and constructively - with Ministers and their officials from the health departments of the UK. The production of single UK-wide consultation from all four Governments signifies the importance of a UK-level approach in tackling rare diseases.

The consultation itself was published on Rare Disease Day 2012 (29th of February) for a statutory 12 week consultation. This was our opportunity to make sure that we as a campaign group made a strong statement about the proposals outlined in the document, supporting initiatives that were good and useful, while also making clear recommendations on areas that were weak, missing a point or altogether absent from the document. We issued four comprehensive responses to the consultation, one for each home nation of the UK. Our responses were informed by the recommendations from our 'Improving Lives' report and through our interactions with stakeholders and members.

We also took the opportunity to make sure that as many responses as possible were made to the consultation, and in the spring of 2012 we embarked on a series of activities dedicated to raising awareness of the consultation and the opportunity it provided to enable people have their voice heard on issues around rare diseases. More about our work and our activities can be read on page six of this report.

Around 350 responses were submitted to the consultation from organisations and individuals. This is an excellent number and demonstrates that rare diseases should be seen as a major health policy issue. The UK's four health departments made the responses received publically available in the autumn, and our analysis shows that a substantial volume of the responses speak with a similar view to change and improve the current situation. RDUK is in discussions with the Department of Health about progress in developing the final UK National Plan for Rare Diseases following the public consultation in the spring of 2012. It is hoped that the National Plan for Rare Diseases will be launched before summer 2013.

The consultation for the National Plan for Rare Diseases

Alongside the 12 week consultation period on the National Plan for Rare Diseases in the spring of 2012, we held a series of activities to support our members and all of the rare disease community across the UK in making their own consultation response.

The activities included the delivery of four one-day events in each of the home nations attended by over 250 people. The attendees included 112 patients or patient organisation representatives, 40 clinicians/healthcare professionals, 49 representatives from industry and 22 academics, amongst others. The aim of the stakeholder engagement events was to debate the issues raised and share viewpoints to encourage our members and other stakeholders to think about how the consultation relates to them, and in turn to gather stakeholders' views to inform our own responses. RDUK believed that in order to get the UK's health departments to take the issue of rare diseases seriously, and to influence a National Plan which will improve access to treatment and care for those with rare diseases, both the quantity and quality of the responses received by the health departments was important. The events were designed to encourage stakeholders to respond, discuss issues with others and to hear expert opinions, helping them to consider what they would like to see in the National Plan.

Our evaluation of the events showed that 100% of those who gave feedback agreed that the stakeholder engagement events helped them to better understand the details of the rare disease plan under consultation. Several respondents said that they were not initially intending to respond to the consultation but after attending the event, each one stated that they decided to submit a response.

'[The stakeholder engagement event] has been extremely useful to help sort the important issues from the chaff. I appreciated the wide range of experiences in my discussion group'
– The Haemophilia Society.

'This was very useful, not only in helping me to write a response to the consultation but also as a networking occasion. [I] thoroughly enjoyed myself and took down masses of notes. Many thanks to RDUK for this opportunity and for leading the way.'
– Trustee, Batten Disease Family Association.

- 4 webinars, viewed 400 times
- Briefing documents, downloaded 1,346 times
- 4 events across the UK, attended by 250 people

Alongside the events, we produced four freely available webinars tailored to each home nation; whereas the aim of the engagement events was to encourage attendees to consider the issues arising in the consultation document independently, the webinars outlined RDUK's views on the consultation. As a result, the two strands of activity complemented each other well. In total 400 people viewed the webinars online and additionally the slides were downloaded nearly 500 times. Our briefing documents to support people in responding to the consultation were downloaded 1,346 times.

Attendees at the London Stakeholder Engagement Event, listening to Lesley Greene from CLIMB discussing Patient care, information and support.

Three other events were held in Belfast, Cardiff and Edinburgh.

'Thank you so much for presenting all the areas of the consultation document with such clarity and insight. One of the real difficulties for the layman is to grasp the key issues in such a detailed document and I really do appreciate your guidance as to how I might frame my response.'
– Ann Price, HAE UK on the support we provided to help organisations respond to the consultation on the Rare Disease Plan.

A growing membership

Interest has risen in and around rare diseases a great deal since our establishment in 2008. We have come a long way in building relationships and increasing support with a great deal of individuals, groups and organisations to become a leading voice in the rare disease community. As of December 2012, our membership stands at over 1,300 and over 230 of these are patient organisations. We are proud of our membership numbers and we will continue to build on this to make sure that the voice of those concerned with rare diseases is heard.

- 1300 members and counting

RDUK across the UK

Northern Ireland

In Northern Ireland, we are making excellent progress through working alongside other groups to form the Northern Ireland Rare Disease Partnership (NIRDP). We are proudly working alongside our partners: the Patient Client Council, with other Health and Social Care Agencies in Northern Ireland, the Irish Platform for Patients' Organisations Science & Industry (IPPOSI) and the Genetic and Rare Disorders Organisation (GRDO), in the Republic of Ireland. By working together this way, we are building a strong and collaborative rare disease community and are ensuring our campaign is relevant to the needs of those living in NI.

The official launch of the NIRDP was in February 2012 and the Minister for Health, Social Services and Public Safety, Edwin Poots MLA launched the public consultation on a National Plan for Rare Diseases. Many MLAs attended and met with their families, and excellent feedback was received.

'At its meeting on 29 February the Committee for Health, Social Services and Public Safety discussed the Rare Disease event that some of the members attended in the Pavilion, Stormont that morning to celebrate Rare Disease Day. The Committee agreed to write to you to express its congratulations on the event and to commend you on the good work being carried out by Rare Disease UK.'

– A letter from the Northern Ireland Assembly Health, Social Services and Public Safety Committee to the member groups of the Northern Ireland Rare Disease Partnership.

Scotland

A highlight of our activity in Scotland has been our work around access to medicines. In 2011, we submitted a public petition on access to medicines for orphan diseases to the Public Petitions Committee of the Scottish Parliament. Petition 'PE1398' called on the Scottish Parliament to urge the Scottish Government to review the mechanism and methodology used by the Scottish Medicines Consortium to appraise the value of medicines for orphan diseases and to instruct the Chief Medical Officer to revise the criteria for accessing Individual Patient Treatment Requests in relation to orphan diseases. The petition was presented to the Committee, and it took further evidence from other stakeholders including the Association of the British Pharmaceutical Industry (ABPI), the Scottish Medicines Consortium, NHS Scotland Health Boards and the Scottish Government.

The Committee acknowledged the concerns raised in petition 'PE1398' and referred it to the Health and Sport Committee for further consideration of the issues raised. RDUK was invited to give evidence in 2012. The Committee agreed to examine general issues regarding the approval process for newly licensed medicines and the system of Individual Patient Treatment Requests and conducted two oral evidence sessions with representatives from the Scottish Medicines Consortium, ABPI, Royal College of Physicians (Consultants) and NHS Boards, the Scottish Cancer Research Network, Scottish Melanoma Group, Cancer Research UK and the James Whale Fund for Kidney Cancer. In 2013, the Committee will review the evidence it has taken and consider how to conclude this work.

It is hoped that the work of the Health and Sport Committee in this area will be acknowledged by the current Scottish Government review into access to medicines on the NHS, which was launched by Health Secretary Alex Neil in November 2012.

RDUK met with Nicola Sturgeon MSP (the then Cabinet Secretary for Health, Wellbeing and Cities Strategy) to discuss the implementation of the National Plan for Rare Diseases in Scotland and have continued to engage with key policy makers and stakeholders in Scotland. This has included meeting with the Scottish Government to discuss the Strategy for Rare Diseases and how this will be implemented in Scotland. There have also been meetings with NHS Scotland National Services Division, ABPI Scottish Access and Value Group and various patient organisations representing rare diseases in Scotland.

Wales

We continue working closely with Assembly Members at the National Assembly for Wales from all political parties and a key activity during this reporting period was our short debate on rare diseases at the National Assembly for Wales. The debate, held by Rebecca Evans AM, discussed the need to commit to developing a National Plan for Rare Diseases in Wales, complete with timescales for implementation and outcome measures, the delivery of the National Plan to be under the remit of a specified team within the Welsh Government and for a national lead for rare diseases. The Minister affirmed the Welsh Government's commitment to improving services in Wales, and looks forward to the completion of the work on developing a National Plan for Rare Diseases following the public consultation. The Minister promised to update AMs in due course about the way forward.

We were invited to provide a briefing about access to medicines for orphan medicines from the patient perspective at the National Assembly's Health and Social Care committee's briefing seminar for committee members to gain a greater understanding of the issue.

We also submitted several consultation responses during this period. They included the public consultation on the all-Wales decision making policy on Individual Patient Funding Requests (IPFR) published by NHS Wales in July 2011. Following the publication of the revised IPFR policy, we submitted a further written response seeking clarity around the criterion of patient exceptionality, and also on monitoring the delivery of the policy. We submitted a response to the Welsh Government's 'Together Against Cancer Plan' consultation.

Research & development, building networks and support for rare diseases

Our efforts this year also extended to improving and strengthening our networks and relationships with researchers across the UK. In the summer of 2012, we collaborated with the National Institute of Healthcare Research (NIHR)'s National Office for Clinical Research Infrastructure (NOCRI) to hold a Rare Disease Research Showcase event in Cambridge. The event brought together key stakeholders including the research community, patients and patient organisations and representatives of the pharmaceutical industry to promote the importance of research into rare diseases and to learn about some of the world-class research into rare diseases taking place at the NIHR's Biomedical Research Centres (BRCs) and Biomedical Research Units (BRUs). The event was sponsored by Professor Willem Ouwehand, University of Cambridge, the UK's lead on the International Rare Disease Research Consortium and Professor John Bradley, Director of the Cambridge BRC.

Following on from this event with the NIHR, RDUK was delighted to attend 'Rare Diseases in the UK: Vision 2020' which brought together influential figures in the research world to discuss issues around rare disease research. Our Chair, Alastair Kent spoke at this event and attendees included Professor Sir John Bell, Academy of Medical Sciences; Professor Sir Leszek Borysiewicz, University of Cambridge; Professor Sir John Savill, Medical Research Council; and Dame Sally Davies, Chief Medical Officer and Chief Scientific Advisor for England. The event focussed on three themes 'Rare Diseases, a Common Problem', 'Building Infrastructure for Rare Disease Research' and 'Improving the Care of Rare Diseases in the UK'. It was pleasing to see rare disease research being given such a high priority and we continue to develop our links with those in attendance.

Financial information

Funding

RDUK is funded by an unrestricted educational grant from its industry members. This funding enables us to offer free membership to all of the other members including individuals, families, groups and organisations.

We are extremely grateful to all of our funders, without which we would not have been able to conduct our activities.

2011

From 1st January - 31st December 2011, RDUK received a block grant of £50,000 from the Orphan Medicines Industry Group (OMIG) within the Association of the British Pharmaceutical Industry (ABPI) and through individual subscriptions of £7,000 each from other non-OMIG member companies, who we informally termed OMIG Partnership. 2011 industry members are listed below.

OMIG Members:

Actelion
Alexion
Amgen
Celgene
GlaxoSmithKline
Merck Sharp and Dohme
Pfizer
Takeda
UCB

OMIG Partnership:

Bayer
BioMarin
CSL Behring
Genzyme
Sigma Tau
Shire

2012

From 1st January - 31st December 2012, RDUK received funding directly from industry members. Each company paid £7,750. 2012 industry members are listed below.

Actelion
Alexion
Baxter
BioMarin
CSL Behring
Genzyme
GlaxoSmithKline
Merck Sharp and Dohme
Novartis
Pfizer
Shire
Sigma Tau
UCB

We also received an additional grant of £7,500 from Novartis to support our work in Scotland and Wales; and we received charitable donations from individuals totalling £8,490.

Financial statement

1st July 2011 to 31st December 2012 (Eighteen months)

Deficit at 1st July 2011 per last statement			£ 20,722
Income			
Membership Fees	£ 143,250		
Donations	£ 8,490		
Total Income		£ 151,740	
Expenditure			
Salaries (inc NI & Pensions)	£ 93,306		
Direct costs	£ 14,797		
Overhead	£ 21,311		
Total Expenditure		£ 129,414	
Surplus in period			£ 22,326
Surplus at 31 December 2012			£ 1,604

During 2012 we also received a total of £29,750 to conduct activities to raise awareness and increase engagement on the consultation on the National Plan for Rare Diseases. The companies who supported this are listed below:

Actelion
Alexion
Baxter
BioMarin
CSL Behring
Genzyme
GlaxoSmithKline
Nordic
Orphan Europe
Pfizer
Shire
Swedish Orphan Biovitrum

Rare Disease UK's accounts are independently audited as part of Genetic Alliance UK's accounts running from 1st April 2011 – 31st March 2012, and will be audited from 1st April 2012 – 31st March 2013.

Looking ahead to 2013

The year ahead looks to be busier than ever, as the deadline for publishing the National Plan for Rare Disease is the end of the year. We will continue to work alongside officials and Ministers across the UK governments to ensure this is delivered in good time.

Once the National Plan itself is published, we can look to the real work starting; its effective implementation. We will be campaigning to make sure the structures are in place to deliver it across the four home nations of the UK, including integration on a UK level where necessary, and ensuring outcomes are measured with clear accountability for its delivery.

We will continue to campaign on other features necessary to improve life for people with rare diseases beyond the National Plan, particularly on initiatives around research and development, as well as other policy matters which have a direct impact on rare diseases.

We will work closely with families to achieve media coverage about their condition. We will also stage a EUROPLAN conference to discuss how the National Plan should be implemented across the UK.

We will be launching a series of reports at our Rare Disease Day parliamentary events in February 2013. In Scotland and Wales we will publish our surveys of the experiences of families living in the devolved nations, as well as a report on public and major charitable funding of medical research into rare diseases and a report on the value to patients and the health service of care coordinators for rare diseases.

In 2013 we will also be reviewing how centres of excellence for rare diseases would work in the context of the UK's health systems and working with the Royal College of General Practitioners (RCGP) on their rare disease priority area, particularly looking at issues around improving diagnosis.

2013 looks to be another busy year for the whole team. We are looking forward to continuing our work with our members, as well as forging relationships with new groups and organisations to bring about lasting change offering better health and quality of life for individuals and families affected by rare diseases.

About Rare Disease UK

Rare Disease UK (RDUK) is the national alliance for people with rare diseases and all who support them. Membership is open to all and includes patients and family members living with a rare disease, patient organisations, clinicians, researchers, academics, and industry. RDUK provides a unified voice for the rare disease community, capturing the experiences of patients and families and raising the profile of rare diseases across the UK.

RDUK was established in 2008 following the European Commission's Communication on Rare Diseases: Europe's Challenges. Following this Communication, RDUK successfully campaigned for the adoption of the Council of the European Union's Recommendation on an action in the field of rare diseases (June 2009). The Recommendation outlines how EU member states should develop and implement plans or strategies for rare diseases by the end of 2013.

Since the adoption of the Council of the European Union's Recommendation, RDUK has been actively ensuring that the UK's health departments act on their obligation to develop a National Plan for Rare Diseases and we have been instrumental in engaging the rare disease community to shape its content.

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Genetic Alliance UK
Supporting. Campaigning. Uniting.

Rare Disease UK is an initiative of Genetic Alliance UK, the national charity of patient organisations supporting all those affected by genetic disorders.

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