

RARE DISEASE | UK

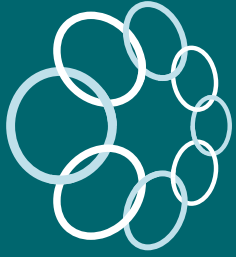
Annual Report

July 2009
June 2010



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

www.raredisease.org.uk



RARE DISEASE | 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 campaigning for a strategy for integrated service delivery for rare diseases. This would coordinate:

- research
- prevention and diagnosis
- treatment and care
- 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.

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Chair's Summary

Reading this Annual Report cannot help but demonstrate that we have made considerable progress during what has been an eventful year. Twelve months ago we were still working to convince policy makers and politicians of the need for a coordinated strategy for the provision of services and support for families living with rare diseases.

Now the question is not "whether" but "how", and representatives of the four home nations are meeting to coordinate their thoughts as to how this might be achieved. Rare Disease UK is playing a key role in this, inputting ideas to the health departments and the NHSs in all four home nations, and meeting regularly with key officials and politicians in Cardiff, Edinburgh and Belfast as well as in Westminster and Whitehall.

RDUK's message about the need to work smarter, and to make more effective use of the money currently spent by the NHS on all too often poor services for families affected by rare diseases is well received, especially in the current climate where the watchword is effective use of resources.

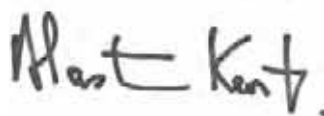
In the middle of all this activity the General Election brought a change of Government, and hard on its heels the announcement of the most significant shake-up in the NHS in England for sixty years.

We were pleased that the importance of commissioning for specialist services has been recognised as a responsibility for the proposed National Commissioning Board, and we will be working hard to ensure

that the new framework properly reflects the needs of patients and families and that it integrates with arrangements elsewhere in the UK. The timescale for implementing the changes that the Government has outlined is a tight one. RDUK will be working with friends and colleagues in order to ensure that the special needs of those with rare diseases are firmly in the foreground when plans are being made and resources allocated.

We look forward to the coming year. It will be a busy and a challenging one, but we have an opportunity to make real progress which we are determined to exploit.

Finally, this report gives me an opportunity to acknowledge the contribution of all those who have played a part in our work – the staff of Genetic Alliance UK, the chairs and members of the Working Groups, the members of the Management Committee and all our members and supporters without whom we could not have achieved any of the progress in this report.



Alastair Kent
Chair of Rare Disease UK

Executive Summary

Rare Disease UK's (RDUK) constitution was formally signed in July 2009. This came on the back of RDUK's success in campaigning for the adoption of the Council of the European Union's *Recommendation on an action in the field of rare diseases* which was adopted unanimously by each EU Member State on the 9th June 2009. The Recommendation highlighted the issues involved in providing effective services to patients with rare diseases and in conducting research.

It was recommended that Member States develop plans or strategies for rare diseases to ensure the integration of services, facilitation of research and ultimately to ensure that patients with rare diseases are able to access high quality care. June also saw the appointment of a dedicated secretariat, Stephen Nutt, to work on Rare Disease UK.

Over the year following the adoption of the Council Recommendation, RDUK sought to engage with the UK Government as well as the Devolved Administrations in Scotland, Wales and Northern Ireland to communicate the need for a strategy for rare diseases and to secure a commitment to developing a strategy. As part of this process we developed relationships with key civil servants and undertook a process of political engagement in all four home nations. As a result of this work we are delighted that there has been a commitment by all four health departments to work together to develop a strategy.

Another priority for the year was to establish and develop RDUK's five Working Groups comprising multi-stakeholder experts. The purpose of the Working Groups is to aid the development of a strategy by identifying issues that need to be addressed and recommendations as to how this might be achieved. RDUK gained its second full-time member of staff, Lauren Limb, during this period and Lauren has been instrumental in coordinating the Groups and building on the outputs of the Groups through additional research.

Looking forward to the year ahead, RDUK will be working towards the publication of the Working Groups' report on a strategy for rare diseases which will be launched at four parliamentary receptions in each of the four home nations to coincide with Rare Disease Day 2011. We will subsequently begin the process of working with the health departments to ensure that the report informs the development of an effective strategy for rare diseases.

“we are delighted that there has been a commitment by all four health departments to work together to develop a strategy”

Putting a strategy for rare diseases on the agenda

When the UK Government signed up to the Council Recommendation on an action in the field of rare diseases in June 2009, the campaign for a strategy for rare diseases received a significant boost. However, Council

Recommendations are not binding and there was therefore a very real possibility that the Government would merely pay lip-service to the Recommendation when it came to reporting back to the European Commission in 2013.

Indeed, shortly before the Recommendation was signed the Labour Government's position was that there were "no plans to set up a national plan" and "the health and social care needs of people living with rare conditions should be met by local health bodies" *.

** In a letter on the 22nd May 2009 from the then Minister of State for Public Health, Dawn Primarolo MP to Adrian Sanders MP*



Melissa Hillier, Stephen Nutt and Alastair Kent outside the Northern Ireland Assembly

An additional threat to the development of a strategy for rare diseases was the uncertainty cast by a forthcoming general election, the timing of which was uncertain. With a change of government likely there was a real danger that the policy would be dropped, especially against a backdrop of cutbacks and efficiency savings.

With this in mind, one of RDUK's main objectives of the year was to communicate the importance of developing a strategy for rare diseases and at the same time to

dispel some frequently held views including:

- there is nothing that can be done to help patients with rare diseases due to the low numbers of people involved
- what can be done would be unfeasibly expensive

We believe that a strategy for rare diseases provides an opportunity to ensure the most efficient use of NHS resources whilst providing better

outcomes for patients. Patients with rare diseases are already heavy consumers of NHS resources, but services are too frequently inefficient or poorly coordinated and patients struggle to access the care, support and treatment they need. Not only does this lead to a reduction in patients' quality of life and potentially life expectancy, it also creates waste through delays in diagnosis, duplication of services, repeat visits to hospital, etc.

RDUK's objective was, and continues to be, to work with the health

departments to ensure that an effective strategy is implemented. Over the year we set out to identify and build links with the key officials in all four governments and NHSs in the UK. We have succeeded in this aim and we now meet regularly with the key figures in all four home nations who have shown themselves willing to cooperate with us as progress is made. We are also pleased to report that officials from each health department are meeting to discuss collaborative working on developing a strategy.

In tandem with our activities in engaging with officials, we also pursued an active campaign of engaging with parliamentarians at Westminster, the Scottish Parliament, Welsh Assembly and the Northern Ireland Assembly to raise awareness of rare diseases and to maintain political pressure to develop a strategy for rare diseases.

We conducted four specific parliamentary contact campaigns over the year:

- Around RDUK's first birthday in November 2009
- In the run-up to Rare Disease Day 2010, both to raise awareness of rare diseases and the Day, but also to encourage attendance at receptions at Holyrood, the Senedd and Stormont
- To prospective parliamentary candidates ahead of the general election to ask for their support on the issue
- To elected MPs following the election to highlight the importance of a strategy for rare diseases and to call on them to join the All Party Parliamentary Group on Rare Diseases

We utilised the support of our members in these campaigns in acknowledgement that constituents have the strongest influence over their representatives. We are very grateful to all our members who participated in these campaigns;

without the support from the strong membership base that RDUK built over the year, we would not have been able to create the same level of impact. One demonstration of the effect of these campaigns was the political impact of Rare Disease Day which is discussed in the next section.

It is testament to the work that RDUK has done over the year that a commitment to developing a strategy for rare diseases has been strengthened, despite a change of government following the general election in May 2010. In a response to a parliamentary question in June 2010, Anne Milton MP, the Parliamentary Undersecretary of State for Health, confirmed for the first time that a strategy for rare diseases is being developed.

The Chief Medical Officer's Annual Report 2009

The campaign for a strategy for rare diseases received a boost in March 2010 with the publication of the Chief Medical Officer (CMO), Sir Liam Donaldson's Annual Report 2009. The Annual Report featured a chapter entitled "Rare is Common" and highlighted many of the issues and messages that RDUK has been working to communicate.

The CMO's report recognised the scale of the issue and that rare diseases have been a largely neglected area of policy. The CMO made a number of recommendations to the Department of Health which are in line with what RDUK is advocating including:

- Strengthening the network of specialist centres for rare diseases
- Ensuring that an adequate number of specialists are trained in rare diseases
- A National Clinical Director to oversee the development of standards and pathways for rare diseases
- National registers to support service planning and delivery
- Strengthening research
- Raising public and professional awareness
- Cross-border collaboration to share information and resources

For the first time a high profile figure in the Department of Health was championing a more systematic approach to addressing service delivery for patients with rare diseases. RDUK has since worked to build on the foundations laid by the CMO.

Available at http://www.dh.gov.uk/en/Publicationsandstatistics/Publications/AnnualReports/DH_113912

"In 2008, the Genetic Interest Group [the former name of Genetic Alliance UK] joined others to create Rare Disease UK. This alliance is working with government, pharmaceutical companies, academics and clinicians to develop a rare disease strategy and to raise public awareness of rare diseases."

RDUK is recognised as an integral organisation in developing policy for rare diseases in the Chief Medical Officer's Annual Report 2009



An opportunity for politicians to meet with their constituents

Rare Disease Day 2010

Rare Disease UK marked Rare Disease Day 2010 by holding parliamentary receptions at the Scottish Parliament, the Welsh Assembly and for the first time the Northern Ireland Assembly. These events saw over 300 attendees from a variety of backgrounds get together to share experiences, raise awareness and demonstrate the need for high-quality services for rare diseases.

"[I would like] to congratulate you and your colleagues for a very effective meeting last night. It was really well done and worked very well. I am sure that everyone made new contacts. It was a pleasure to take part."

Professor Sir Ian Wilmot, who spoke at the Scottish Parliament event

Many attendees and members had participated in our contact campaign leading up to the event and we secured an impressive turnout of politicians including 17 MLAs in Stormont, at least 10 MSPs in Holyrood and 19 AMs in the Senedd (and a further 7 contacted us saying they would have attended had the plenary session not overrun). We received excellent feedback on the events from all stakeholders, including politicians.

We did not hold a reception at Westminster this year due to the timing of the general election. Nevertheless RDUK and our members were active in engaging with MPs to inform them of the issues affecting people with rare diseases. An Early Day Motion



Chair of RDUK, Alastair Kent speaks to Lord Browne MLA, co-host of the event at Stormont



Chief Medical Officer, Dr Tony Jewell addresses the Welsh Assembly event

welcoming Rare Disease Day and calling for a strategy for rare diseases was launched by Dr Evan Harris MP. This was signed by a total of 71 MPs – an impressive number given the short amount of time before the dissolution of parliament and an apparent caution from Conservative MPs. A Motion launched in the Scottish Parliament was signed by 35 MSPs – 14 more than a similar motion the previous year.

RDUK was also active in generating media coverage around Rare Disease Day. With the cooperation of our members who submitted many case studies and the assistance of Velvet PR we generated over 10 published articles and 5 pieces of broadcast coverage.

“I should like to thank you for all the hard work and preparation you did for Rare Disease Day at the Northern Ireland Assembly. The reception was very well attended and has generated considerable interest and discussion. I have spoken to many people who attended and they all agreed it was very worthwhile and well organised.”

*The Lord Browne of Belmont
MLA on the reception
at Stormont*

One very positive example of the effect of these media stories was when the Welsh Assembly Government finally granted confirmation to patients with Laurence Moon Bardet-Biedl Syndrome (LMBBS) that they would be funded to access a new National Commissioning Group service in England. Following a two page spread that we secured in the Western



Professor Sir Ian Wilmut speaks at the Scottish Parliament

Mail (Wales’ most read national newspaper), pressure was put on the Government to announce their decision regarding funding, a favourable one which will make

“...the evening was very, very beneficial to us and we chatted to a couple of AMs and [other key stakeholders] which proved to be very interesting and informative on both sides. I hope that in talking to the people that we did we raised awareness to the ministers of what as a society we are about and what we would like to do in the future with their help.”

*Hayley Cleaver, Chair,
Turner Syndrome Support
Society on the event at the
Welsh Assembly*

a tremendous difference to the health and well-being of patients with this rare disease.

Rare Disease Day activities in the UK coincided with events taking place across Europe and internationally. RDUK is actively involved in Rare Disease Day on a wider scale through our role on the Eurordis (the European Organisation for Rare Diseases) Council of National Alliances.

A full report on all Rare Disease Day activity is available on the RDUK website.

Working Groups

One of RDUK's priorities over the year was the establishment of five Working Groups to inform a report into a strategy for rare diseases. The Working Groups have been gathering evidence of the issues currently facing patients and families with rare diseases, what an effective strategy needs to implement to address these issues, and the principles that must underline the strategy.

The five Working Groups comprise a total of approximately 60 experts from all the stakeholder groups with an interest in rare diseases.

The Groups are:

Coordination of Research

Chaired by Dr Hilary Burton, Programme Director – Foundation for Genomics and Population Health (PHG Foundation)

Prevention & Diagnosis

Chaired by Dr Jim Bonham, Laboratory Director of Newborn Screening, Sheffield Children's NHS Foundation Trust

Commissioning and Planning

Chaired by Adrian Pollitt OBE, former Director of National Specialised Commissioning

Patient Care, Information & Support

Co-chaired by Andrew Wilson Webb, CEO - Rarer Cancers Forum and Lesley Greene – Vice-President, Climb and Eurordis Patient Representative

Delivering Co-ordinated Care

– Dr Atul Mehta, Consultant Haematologist – Royal Free Hospital

Lauren Limb was employed in March 2010 as RDUK's second full-time member of staff. Lauren has been instrumental in developing the Working Groups. As well as coordinating the Groups, Lauren has been conducting research to build upon the outputs from the Working Group meetings as well as gathering examples of good and bad practice which can inform the development of a strategy.

In the summer RDUK launched its survey of patients' and families' experiences of rare diseases which will inform the Working Groups. The outcomes will also be invaluable in supporting our communications activities.

The initial findings from the Working Groups will go out for consultation to our members and a wider base of stakeholders in the autumn of 2010. The final report of the Groups will be published and launched to coincide with Rare Disease Day 2011.





Other achievements

We gave presentations about RDUK at the following conferences/ events

- Alpha 1 Awareness UK Patient Information Day (September 2009)
- All Wales Immunodeficiency Patient Evening (November 2009)
- UKPIN/British Society for Immunology Conference (November 2009)
- European Genetic Alliances Network meeting in Basel (January 2010)
- Rare Disease Day reception at Belfast City Hall (February 2010)
- ALD Life Patient Day (May 2010)

We exhibited about RDUK at the following conferences:

- European Paediatric Neurology Society Rare Disorders Symposium (September 2009)
- British Paediatric Neurology Association Annual Conference (January 2010)
- Sickle Cell: The Next 100 Years Conference (April 2010)
- Association of Genetic Nurses and Councillors Annual Conference (April 2010)
- Ability NI Disability Exhibition 2010 (June 2010)

Consultations responded to:

- House of Commons Health Committee Inquiry into Commissioning
- Department of Health Consultation on the Innovation Pass Pilot
- Department of Health Consultation on Strengthening Specialised Commissioning
- Academy of Medical Sciences' review of the regulation and governance of medical research

- Doubled the membership to approximately 500 members including over 120 patient organisations.
- The number of hits on the RDUK website almost doubled from 33,350 in September 2009 to 63,906 in June 2010
- Produced new promotional material.

- Hosted a fringe debate at the Conservative Party conference in October 2009. Earl Howe who hosted the event is now part of the Department of Health Ministerial team.
- An afternoon of workshops dedicated to Rare Disease UK was held at the Genetic Alliance UK Annual Conference

- We worked with the All Party Parliamentary Group on Rare Diseases. The Group was chaired by Anne Milton MP who was appointed to the Department of Health Ministerial team following May's general election. Since the election we have been working to re-establish the Group.

Financial Information

January 2009 – December 2010

Income			
Donations / Membership fees	£83,000		
		£83,000	
Total Income			£83,000
Expenditure			
Salary	£30,521		
Expenditure	£28,087		
		£58,608	
Total Expenditure		£58,608	
INCOME LESS EXPENDITURE			£24,392
Overheads	£24,416		
Less overheads		£24,416	
Remaining balance @ 31/12/2009			-£24

Projected Financial Information

Jan 2010-December 2010

Income			
Donations/ Membership fees	£98,113		
		£98,113	
Total Income			£98,113
Expenditure			
Salary	£63,669		
Expenditure	£11,832		
		£75,501	
Total Expenditure		£75,501	
INCOME LESS EXPENDITURE			£22,612
Overheads	£22,284		
Less overheads		£22,284	
Remaining balance @ 31/03/2010			£328

Please note: the Expenditure is only up until March 2010. Salaries and overheads are calculated until the end of the reporting year. The forecasted overspend will be met by Genetic Alliance UK.

The Rare Disease UK accounts will be independently audited as part of the Genetic Alliance UK accounts running from 1st April 2009-31st March 2010.

Funding

RDUK is funded by an unrestricted educational grant from its pharmaceutical industry members. Each company, regardless of size, pays an annual subscription fee of £7000, to reflect the fact that each company has equal weighting.

At the beginning of the year, RDUK was mainly funded by the Association of the British Pharmaceutical Industry's (ABPI) Orphan Diseases Industry Group (ODIG). ODIG is a group of orphan drug manufacturers working together to ensure the NHS provides patients with medicines they need. RDUK was also funded by Genzyme who fell outside the ABPI.

In recognition that there were other non-ABPI companies with an interest in supporting the development of a strategy for rare diseases, the Orphan Diseases Industry Group Partnership (ODIGP) was established in July 2009 as a mechanism to allow such companies to input to RDUK.

A representative from ODIG and a representative from ODIGP both sit on the RDUK Management Committee.

During the year RDUK was successful in attracting a greater number of industry members which enabled us to carry out a wider range of activities.

At the end of the year
RDUK had 13 member companies.

ODIG

- Actelion
- Alexion
- Baxter
- Celgene
- Merck Serono
- Merck Sharp Dohme
- Pfizer
- Takeda
- USB

ODIGP

- CSL Behring
- Genzyme
- Shire
- Sigma Tau





Looking Ahead July 2010 - June 2011

RDUK has built excellent networks with key civil servants, gained the respect and trust of important parliamentary figures and brought together a world class group of patients, clinicians, researchers and industry supporters.

With this in mind, the focus for the remainder of 2010 will be to develop the report into a strategy for rare diseases. This will involve the Working Groups continuing to meet, further research being conducted as well as a consultation on our initial findings to allow our members and other stakeholders to respond. Specific events will also be taking place to gather further evidence, including our EUROPLAN conference in November and focus groups in Scotland, Wales and Northern Ireland to highlight specific issues in the devolved nations.

2011 will be a crucial year in the campaign for a national strategy and Rare Disease UK will be instrumental in providing the necessary oversight and scrutiny of the development and implementation of an effective strategy. The NHS in England will be undergoing one of the biggest

reorganisations in its history and this will generate many opportunities as well as potential threats for the development and implementation of a national strategy for rare diseases. We must use the reorganisation of the health service to our advantage in ensuring that our recommendations to improve the lives of those affected by rare diseases are listened to and incorporated into the newly forming institutional structure

The Working Group report will be published to coincide with Rare Disease Day 2011 and the report will be launched at four parliamentary receptions at Westminster, Holyrood, the Senedd and Stormont.

RDUK will then enter the third stage of its campaign and will focus much of its efforts on working with policy makers to ensure the report's messages and findings are built upon by the Department of Health in developing a strategy for rare diseases.

Whilst this is an exciting time it is also crucial that rhetoric is turned into action and that a national strategy begins to be developed. RDUK is leading this campaign in the UK. Without our continued networking, knowledge and passion there is a real risk that the national strategy for rare diseases will not be fully developed or implemented.

"...it is also crucial that rhetoric is turned into action and that a national strategy begins to be developed"

Acknowledgements

We would like to express our gratitude to all those who have supported the work of Rare Disease UK over the past year.

We would particularly like to thank all of our members who have helped us in raising awareness of the importance of a strategy for rare diseases by attending events, participating in our contact campaigns, distributing information through their networks and in many other ways.

Special gratitude also goes to:

The Management Committee

Mark Barrett

Dr Peter Corry

Laura Gilbert

Marie McGill

Dr Stephen Jolles

Dr Marita Pohlschmidt

Steve Potter

Becky Purvis

Dr Fiona Stewart

Also to Julie Ann Bridge who stood down earlier this year.

And Advocate in their role as advisors.

Working Groups Chairs

Dr Hilary Burton

Dr Jim Bonham

Adrian Pollitt OBE

Lesley Greene

Andrew Wilson-Webb

Dr Atul Mehta

Also to all the members of the Working Groups.

The Orphan Diseases Industry Group and to members of the Orphan Diseases Industry Group Partnership





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