



RARE DISEASE | UK

Funding Support for Rare Disease Research:

Raising Awareness
and Increasing
Transparency



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

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 150 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.

RDUK is funded by an unrestricted educational grant from its industry members.

The Recommendation calls on Member States to adopt plans or strategies for rare diseases by 2013.

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

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



Acknowledgements

We wish to express our gratitude to the patients, families, researchers, clinicians, patient organisations and other healthcare professionals who took the time to respond to the survey.

We would also like to thank the researchers and representative from funding bodies who took part in the interviews.

The Rare Disease UK team

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Chair's Foreword

Despite recent advances in our understanding, and the development of a growing number of specific therapies for individual rare diseases, it remains the case that most of the 6000 or so identified rare diseases remain at best intractable. For many we still have a poor understanding of the biology and a number continue to limit the quality and quantity of life for those affected significantly.

For many patients and families living with these conditions, sustainable high quality biomedical research and development offers hope. Hope that there will be something that will change the prognosis, and hope that the continued interest in "their" condition from the research community will lead to greater understanding and a better opportunity to gain insight into the disease and its consequences.

High quality research needs sustainable funding from a range of different sources. In this report we concentrate on some of the major public and charitable sources, seeking to establish the extent to which patients, families and the academic and clinical communities are aware of the opportunities that exist, and the ease (or otherwise) with which these can be accessed. The findings reveal a gap in perception between funders and applicants. The extent to which this gap in perception is based in an objective reality is arguable, and establishing the truth of the situation is beyond the scope of this report. However, to the extent that perception, even if inaccurate, colours the action we take, there is clearly an issue which needs to be addressed. If patients, families and the research community think that there is little opportunity to secure funding for rare disease research, and is difficult to secure, then this can be a significant disincentive. There is a danger that this will create a downward spiral, where applications are few, seemingly indicating a lack of interest in the issue which, in turn reduces the available budget and so reducing the likelihood that researchers will see a sustainable career to be had in rare disease research and reducing the range and quality of applications for funders to support.

Clearly there is always scope for increasing the absolute amount of funding for rare disease research. But, as this report shows, there is also a need to increase awareness, and to encourage a greater dialogue between funders and other stakeholders so that high quality projects can receive support, and patients benefit from the new knowledge that will be generated sooner rather than later. Ultimately we need to see rare diseases moving more quickly from being incurable to treatable and then to either curable or preventable. This possibility will not only benefit patients and families, it will also stimulate the research community and contribute to creating a sustainable environment for research and development that will help lift the threat from rare diseases faced by so many families today.

Thanks are due to all who contributed to this report. We are grateful to you all for being willing to share your insights and your time with us.



Alastair Kent OBE
Chair, [Rare Disease UK](#)
Director, Genetic Alliance UK



Executive Summary

This report aims to capture how the rare disease community perceives the funding support that public and major charitable funding bodies give to research into rare diseases. We have focused on the funding bodies Cancer Research UK, the Medical Research Council (MRC), the National Institute for Health Research (NIHR) and the Wellcome Trust. Through a survey to the Rare Disease UK membership, and a series of interviews with the funding bodies and the researchers, this report highlights the barriers to research and identifies areas of current good practice.

Overall, there is a perception amongst the rare disease community that public and major charitable funding bodies do not support research into rare diseases very well, in particular that there is not enough funding awarded. However, this report also reveals that there is a low level of awareness amongst the rare disease community of the opportunities for research, and feedback was that funding opportunities are not promoted effectively. There is a perception that funding this type of research is left to charities supporting rare diseases. Where these charities do exist they are small, often volunteer-run, disease specific and have no, or very limited capacity for funding research and for many rare diseases, there is no specific charity who could fund research.

Rare Disease UK believes that this perception is not entirely true; public and major funding bodies do fund research into rare diseases, many of them have funding streams or awards specifically to encourage research into rare diseases. However, it is difficult to get a clear picture of exactly how much funding is made by these bodies as it is not recorded in an easily accessible or comparative way.

Across all the funding bodies we looked at, there are examples of good practice, however these need to be better promoted to the rare disease community. The perception of poor support for rare diseases can be tackled by helping patients, researchers, healthcare professionals and patient support groups to be aware of rare disease funding streams and awards. The Research Support Service¹ provides excellent advice and information through the process of designing research studies and submitting proposals; however this service is not promoted enough amongst the rare disease research community. There is also an opportunity to better promote and support current funding in rare diseases, to raise the profile of the research that is already taking place.

This report outlines recommendations for improving the perceptions of public and major charitable funding bodies amongst the rare disease community. They are aimed at all four of the funding bodies that this report focuses on but are also good indicators for all funding bodies working within this field of research. Many of these recommendations are easily implemented, but could vastly improve the support that funding bodies give to research into rare diseases.

1. <http://www.ccf.nihr.ac.uk/Pages/RDSMAP.aspx> - The NIHR Research Design Service (RDS) network provides help for researchers to prepare proposals for submission to NIHR and other national, peer-reviewed funding competitions for applied health or social care research by providing expert advice on research design and methodology.

Recommendations

Recommendation 1: Awareness needs to be raised of funding opportunities for rare disease research through better promotion of existing programmes.

Recommendation 2: Monitoring what is spent on rare disease to enable funding bodies to identify the proportion of their resources allocated to rare disease research.

Recommendation 3: Methods to foster collaboration in the field of rare disease research should continue to be explored and exploited.

Recommendation 4: Initiatives such as Research Design Service should be better promoted to support researchers in the field of rare diseases.

Recommendation 5: Peer review of funding applications should include someone with experience in rare disease research.

Additional Findings

Patient Registries and NHS Research and Development Approval

Although not within the scope of our recommendations, two additional areas of concern were raised in the survey responses. One of the main issues both patients and researchers raised was the lack of infrastructure for rare disease research. It is often difficult to identify patients eligible to take part in research and this is due to the uncoordinated manner in which data is collected, stored and shared.

Many patients and families recognise the first step in research as gathering a registry for their condition in order to collect a pool of data which can then be used for research. However, respondents indicated that they faced barriers to long term funding of registries and researchers noted that the manner in which data was collected in the registries that do exist was often inconsistent and therefore not comparable.

Rare Disease UK see registries as a key tool to understanding the complexities of rare disease and one which is currently being significantly under resourced and for which a more consistent approach is required.

The other issue that was raised within the survey responses was that of the NHS research and development approval process within the UK. The very nature of rare disease means that researchers are often collecting data from various sites around the country and many raised concerns about the time taken to gain research and development approval within each recruitment site. Steps have been taken to streamline this process but it is clear from our respondents that there are still discrepancies and additional hurdles within each research site, these include extra forms, variation on forms and the length of time taken to receive approval.

Rare Disease UK values that the research and development process is an important part of research, however we find it frustrating that in an area where research is lacking, burdensome hurdles mean there are often long delays which reduce the resources available.

Introduction

Scientific and medical research is vital for increasing our knowledge on rare diseases. As we learn more about the underlying biology of an illness, we can apply that understanding to developing diagnostic tests, effective therapies and treatments and where possible, take preventative measures that can help individuals and their families who are affected by rare diseases. Research into rare diseases can also give insights into common conditions and health generally.

“Just because something is rare [it] doesn’t mean that it’s not important and can give one very incredible general insights into health and disease.”

Sir Mark Walport, Director, Wellcome Trust²

According to RDUK’s members, patients with rare diseases understand the importance of research and many are eager to be involved in research. Our 2010 report ‘Experiences of Rare Diseases: An insight from Patients and Families’³ highlights how patients feel that there are few opportunities to participate in research and there is little awareness of studies into their condition.

“I always try to keep up to date with my condition online. I am always researching but would love further involvement into research.”

Patient with Ollier’s disease/Maffucci syndrome

“I would be very willing to participate in trials or research.”

Patient with Langerhan’s cell histiocytosis

We often hear from both patient groups and patients that they believe public and major charitable funding fails to support research into rare disease and the limited support that is available comes from small organisations and charities, many of which have limited resources.

The summary of responses to the consultation on a UK rare disease plan⁴ reveals dissatisfaction with the current funding support given to research into rare diseases. Many responses acknowledged the lack of promotion of opportunities for or findings of rare disease research and many called for increased or ‘ring fenced’ funding for research into rare diseases.

The UK health departments’ consultation asked, “Do you agree that registries are an important tool in rare disease and could be a core component of the service specification of an expert centre?” 90% of those that responded to this question agreed that registries were an important tool. Many of these responses highlighted the key role registries played in research.

The aim of this report is to assess how the rare disease community perceives public and major charitable funding support for research into this area, to investigate whether the perception of these stakeholders is

2. The Life Scientific Interview, BBC Radio Four, October 2012, <http://www.bbc.co.uk/programmes/b01n111x>

3. ‘Experiences of Rare Diseases: An Insight from Patients and Families’, Limb et al, Rare Disease UK, December 2010, <http://www.raredisease.org.uk/documents/RDUK-Family-Report.pdf>

4. ‘The Consultation on a UK rare disease plan: Summary of Consultation Responses’, Department of Health, November 2012, <https://www.wp.dh.gov.uk/publications/files/2012/11/Consultation-on-the-United-Kingdom-Plan-for-Rare-Diseases-Summary-of-Consultation-Responses.pdf>

accurate, and to identify whether there are areas where funding could develop their support for research. This has been done through a survey of rare disease stakeholders and interviews with researchers and funding organisations which are both outlined in the methodology section of this report.

Throughout this report we have also highlighted areas of good practice, where public and major charitable funding bodies are taking steps to improve the landscape for rare disease research.

This report focuses on four of the biggest public and charitable funding bodies in the UK: Cancer Research UK, the Medical Research Council (MRC), the National Institute for Health Research (NIHR) and the Wellcome Trust, who combined fund approximately £2.6 billion's worth of medical research a year⁵.

5. The Medical Research Council spent £759.4 million on research in 2011/12, Medical Research Council Fact and Figures, <http://www.mrc.ac.uk/about/factsfigures/index.htm>
Cancer Research UK spent £300 million in 2011/12, 'Cancer Research UK Research Strategy Progress Report 2011/12', March 2012 The Wellcome Trust spend on average £600 million a year, 'The Wellcome Trust - Frequently Asked Questions' <http://www.wellcome.ac.uk/News/Media-office/Fact-file/index.htm>
The NIHR spent £919.5 million in 2009/10, 'Embedding Health Research; National Institute for Health Research Annual Report 2009/10', July 2010

Methodology

The intention of this report is to build on the research carried by RDUK in the development of the report 'Improving Lives, Optimising Resources: A vision for the UK Rare Disease Strategy'⁶ and the perceptions of research into rare diseases revealed in our 'Experiences of Rare Diseases' report. A core part of the research entailed a series of interviews with public and major charitable funding bodies of rare disease research and with individual researchers with a background in rare diseases.

RDUK asked representatives from funding bodies about how they support rare disease research; how they recorded their spending in this area, what they perceive to be the challenges facing research into rare diseases and how they can better support rare diseases in the future.

Additionally, written requests were sent to each funding body, asking how much they spend on research into rare diseases. Please see Annex 5 in this report for further details.

Researchers were asked what they believe to be the challenges facing research into rare diseases and how they think research into rare diseases can be better supported. They were also asked about their experiences of applying for funding bodies for research into rare diseases, whether there were any obstacles in this process and whether they could be better supported.

Full detail of the interview questions and interviewees can be found in annexes 1, 2 and 3.

In order to gauge the perceptions of rare disease stakeholders of how public and major charitable funding bodies support rare disease research, we conducted an online survey. We received over 330 responses to the survey, 54% of whom were patients, 16% were representatives of patient organisations, 19% were researchers and 15% were clinicians. The survey questions can be found in annex 4.

6. 'Improving Lives, Optimising Resources: A Vision the UK Rare Disease Strategy', Rare Disease UK, February 2011

Perceptions of research into rare diseases

Overview

Rare disease stakeholders and RDUK's members often report that public and major charitable funding bodies do not provide sufficient support for research into rare diseases. The intention of this survey was to capture the perceptions of support available for research into rare diseases and to gather further understandings about this perception. As a result, the responses are a reflection on how public and major charitable funding is perceived by those in the rare disease community and may not always be an accurate reflection on how funding bodies support research into rare diseases.

The results revealed rare disease stakeholders perceive rare disease research to be poorly supported by major funding bodies.

"I am not aware of any research into my own condition, myotonia congenita, which is the basis of my opinion."

Patient with myotonia congenita

"There seems little effort to manage serious but rare conditions and research possible cures leaving this largely to the voluntary sector."

Rare disease patient

The responses to the survey highlight how generally respondents thought that these funding bodies did not spend enough on research into rare diseases, did not provide sufficient support to applicants and did not adequately promote funding opportunities. Again, this is a perception of the rare disease community and may not be a true reflection of the current situation.

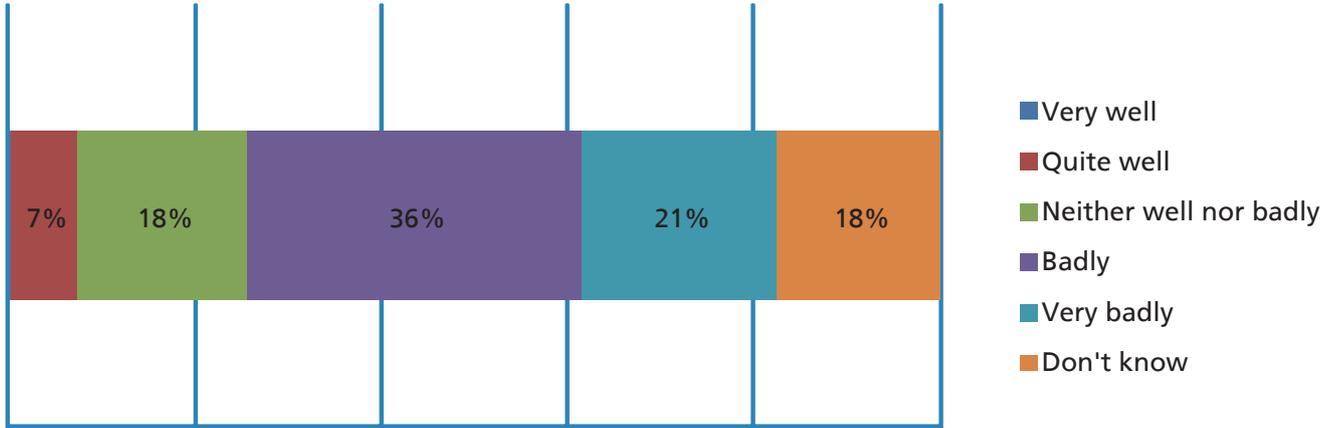
Respondents generally perceived that researchers interested in rare diseases do not encounter a level playing field when submitting proposals for research compared to more common conditions.

Many respondents also felt that opportunities for research were currently not being promoted well enough. Additionally, very few respondents offered feedback on specific funding bodies and many of them responded to questions on specific funding bodies with 'don't know'.

General perceptions

Overall, just over half of stakeholders (57%) said the funding bodies supported research into rare diseases either 'badly' or 'very badly'. Only 0.4% said that the funding bodies supported research very well and only 7% felt that rare disease research was supported 'quite well'.

"How do you feel UK public and major charitable funding bodies support rare disease research?"



Comments made by respondents revealed that they felt rare disease research was neglected by the major funding bodies in favour of more common diseases and that research into rare diseases was left to small charities. Rare diseases charities, where they do exist, are often small, volunteer-led and disease specific and many rare conditions do not have any charity to fund research. Therefore if research is left to this sector, there will be significant gaps and missed opportunities.

"A lot of focus has been on high population diseases supported to a large extent by the extremely well funded charities associated with mass disease. Outside of the rare disease charities, funding from public bodies has been poor over the years although it is beginning to improve."

Rare disease researcher

"While all the organisations below do have some support of rare disease projects, these are not high profile and there does not seem to be a concerted effort to specifically support rarer conditions."

Representative of a funding body/Healthcare professional

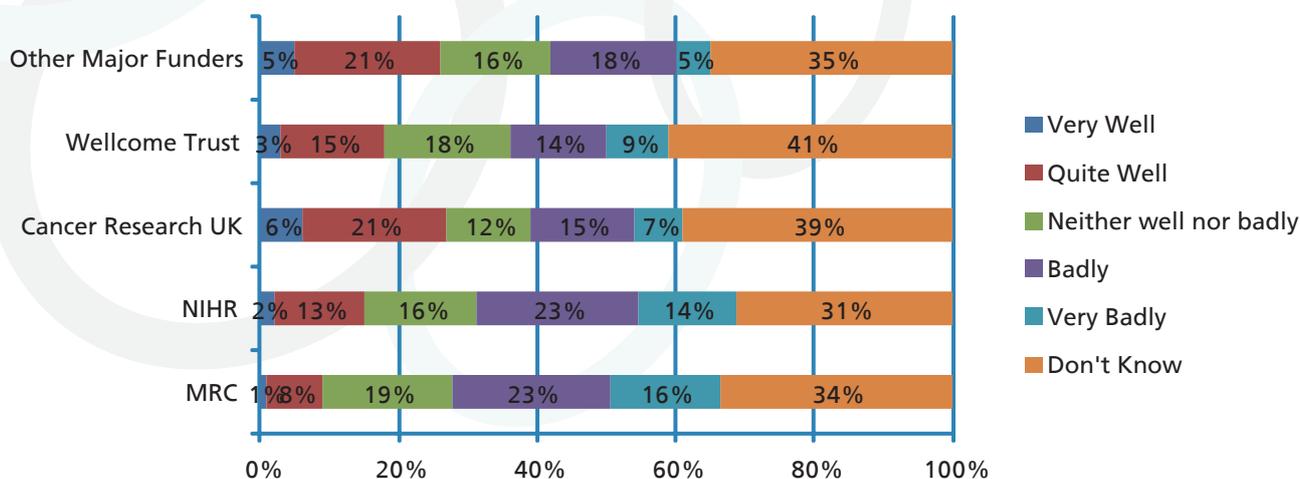
"Often despite scientific excellence of proposals, it is not funded because it is rare."

Clinician

Perceptions of individual funding bodies

When it came to assessing the individual organisations, the majority of respondents said that they didn't know to what extent each of these supported research into rare diseases. Many respondents said that they did not know enough about each organisation's funding of rare disease research to be able to respond. This indicates a significant lack of awareness of how funding bodies support research into rare diseases.

"How do you feel each organisation supports research into rare diseases?"

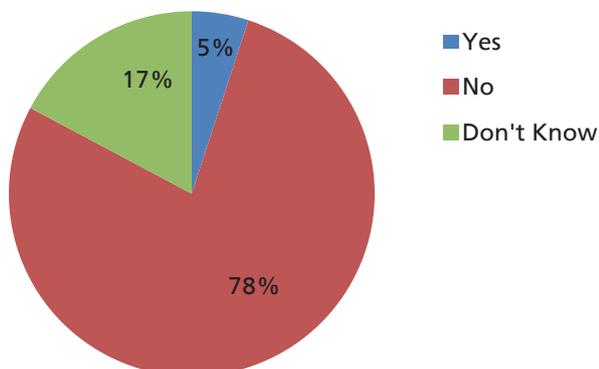


When respondents did express an opinion, most indicated that each funding body generally does not do very well in supporting research into rare diseases.

Availability of funding

The majority of those who responded to our survey (78%) said that the funding bodies did not make enough funding available for research into rare diseases. Less than 10% of respondents (5%) thought there is sufficient funding available.

"Do you think that UK public and major charitable funding bodies make enough funding available for rare disease research?"



"It is very much a case of rare disease organisations having to chase down funding."

Patient

"[The funding opportunities are] usually buried in their websites."

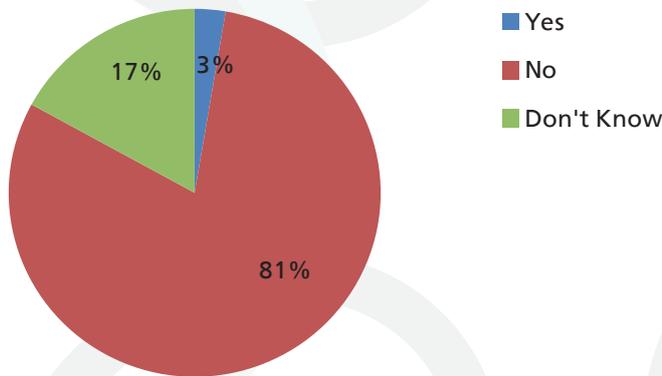
Researcher

"Clearly a deficit that could be addressed. most highlights are on bigger outcome diseases."

Researcher/Clinician

When stakeholders were asked about promotion of funding opportunities for rare diseases, 81% said that they thought that funding bodies did not do enough to make people aware of funding opportunities.

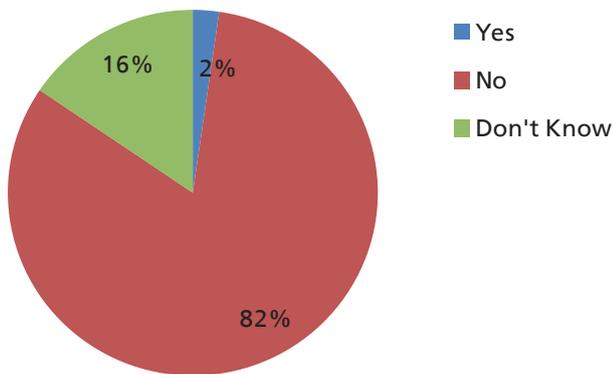
"Do you think that UK public and major charitable funding bodies do enough to make people aware of funding opportunities for rare disease research?"



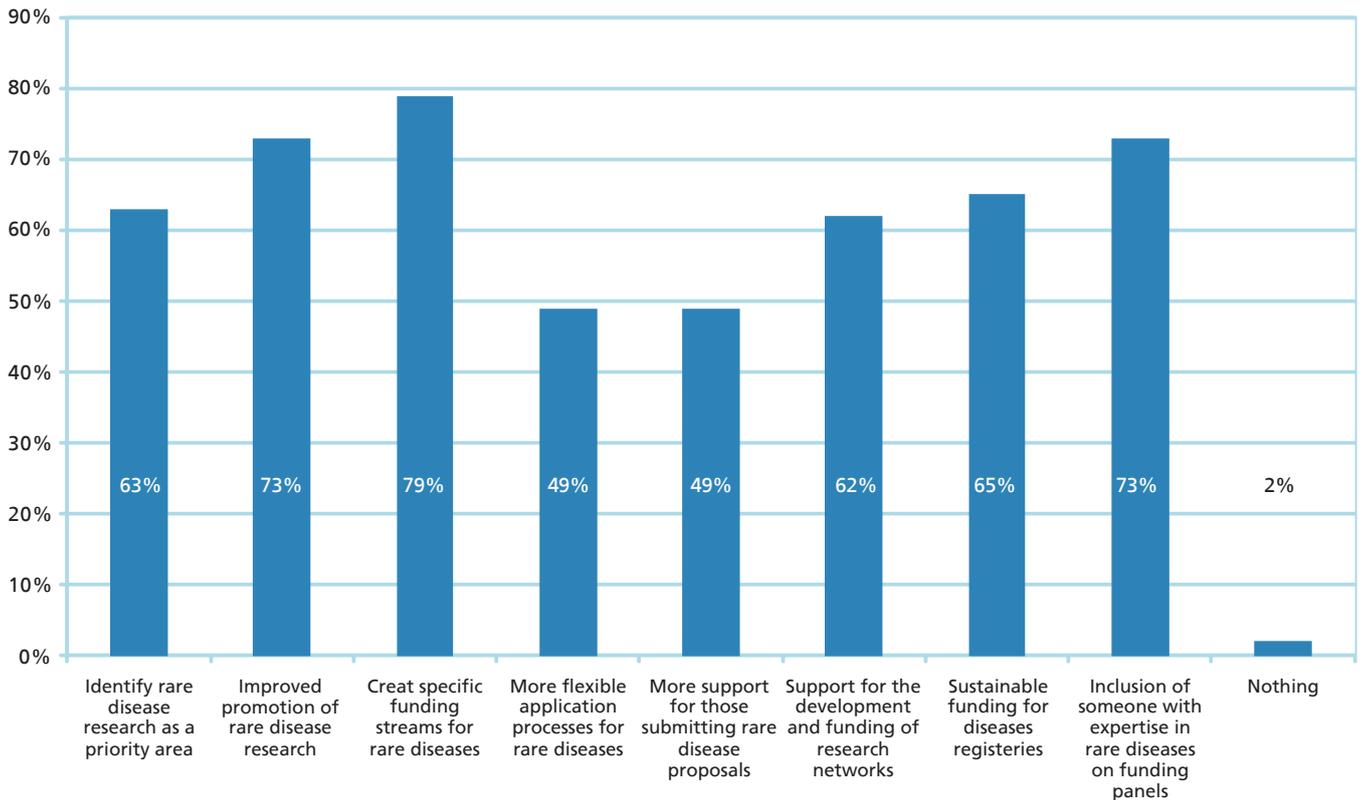
Equality between rare and common diseases

An overwhelming majority of the survey respondents (84%) said that they did not feel there was a level playing field between rare diseases and more common diseases when it comes to applications for funding. Only 2% felt that rare diseases are given the same consideration as more common diseases.

"Do you think there is a level playing field between applications for funding research into rare diseases and research into more common diseases?"



"What do you think UK public and major charitable funding bodies could do to better support rare disease research?"



Stakeholders were asked in the survey what they thought could be done to improve public and major charitable funding bodies support for research into rare diseases. The most popular options amongst stakeholders were to create specific funding streams for rare diseases, including someone with expertise in rare diseases in funding committees and improved promotion of funding opportunities for research into rare diseases.

The survey results largely support what RDUK has been told by rare diseases stakeholders; there is a general feeling that public and major charitable funding bodies neglect rare conditions in favour of more common diseases and that they do not spend enough on rare diseases. However, this is coupled with the feeling that funding bodies do not promote any funding opportunities there are in this area currently and that for the most part stakeholders cannot say how each specific funding body supports rare diseases.

Many participants highlighted how research into rare diseases could also be facilitated by other measures such as patient registries for rare diseases, improved support for researchers submitting proposals, better promotion of funding opportunities and streamlined NHS research & development (R&D) approval and ethical review processes.

Main Findings

Funding

The vast majority of those who took part in our survey said that they felt not enough funding was made available to research into rare diseases. Many stakeholders felt that funding is prioritised for research into diseases that have a greater economic impact.

“By the nature of the rarity of the disease, funding opportunities are restricted.”

Researcher

“A lot of focus has been on high population diseases supported to a large extent by the extremely well funded charities associated with mass disease.”

Researcher

“Unless your research interest is in a ‘big area’ it is hard to get funding.”

Patient

Our survey of stakeholders and our interviews with researchers revealed a perception that public and major charitable funding bodies make decisions about funding based on the prevalence of the condition.

Many of our survey respondents also said that they had not heard of research being funded into their disease areas, or that it was hard to find out about funding opportunities. 81% said that public and major funding bodies don't do enough to make people aware of the opportunities for funding rare disease research. Despite these perceptions, on further investigation it was revealed that Cancer Research UK, the NIHR and the Wellcome Trust all currently have grants, funding streams or themed calls for rare disease areas. The low levels of awareness of these initiatives suggest they are not promoted enough to rare disease stakeholders.

There is a perception amongst stakeholders that funding bodies do not make funding available for research into rare diseases, but from the interviews conducted with the four funders it has found that funding is made available through several schemes, but is perceived to not be promoted to researchers who may apply or the rare disease community who are interested in what is spent on rare diseases.

Often themed calls for research have a lasting impact on the number of funding applications and initiatives into rare diseases. Funding bodies indicated to us that themed calls in other areas of health, such as dementia and stroke have stimulated interest and led to a long term increase in applications. We believe that better promoted themed calls or highlighting notices for rare diseases will help public and major charitable funding bodies communicate the opportunities that are available which in turn will encourage more researchers to take an interest in the area. There is also an opportunity to better promote and support current funding in rare diseases, to raise the profile of the research that is already taking place.

Where they do exist, it is sometimes hard to find information about these themed calls. Opportunities for research into rare diseases should be widely disseminated and promoted, to researchers and through patient organisations.

“It is extremely difficult to identify sources of research funding and you have to rely on experienced fellow organisations to point the way.”

Representative of a patient organisation

“If they are funding it, they should be publicising that funding/support much better.”

Patient

“Usually buried in their websites.”

Representative of a patient organisation

NIHR - Call for proposals Applied Clinical Research on Very Rare Diseases⁷

The NIHR's call for proposals on research into very rare diseases is an example of how to encourage research into rare diseases by promoting existing sources of funding. The call was across six of the NIHR's research programmes:

- Health Technology Assessment (HTA) Programme
- Efficacy and Mechanism Evaluation (EME) Programme
- Research for Patient Benefit (RfPB) Programme
- Health Services and Delivery Research (HSandDR) Programme
- Invention for Innovation (i4i) Programme
- Programme Grants for Applied Research (PGfAR) Programme

The call covers diseases that affect less than 1 in 100,000 of the general population. The call does not set aside funding but aims to attract researchers to the field of rare diseases and highlight the need for research in this area.

Recommendation 1: Awareness needs to be raised of funding opportunities for rare disease research through better promotion of existing funding programmes.

7. <http://www.nihr.ac.uk/research/Pages/VeryRareDiseasesResearch.aspx>

Monitoring spending on rare diseases

Currently public and major funding bodies of research do not monitor their spending on rare diseases in an accessible or comparative way. It is apparent from the interviews we undertook that although funding bodies collected general information it is difficult to ascertain the resources being spent on rare diseases.

Improved monitoring of rare disease spending will allow funding bodies to accurately assess what is spent, whether the expenditure is comparable with that of other disease areas and to ensure spending is effective. If funding bodies can readily identify their spending on research related to rare diseases they can monitor progress in this area and reflect upon the success of themed calls, highlight notices and dedicated funding streams. It would also allow funding bodies to be more transparent about what they fund.

Recommendation 2: Monitoring what is spent on rare disease to enable funding bodies to identify the proportion of their resources allocated to rare disease research.

Collaboration

RDUK's 'Improving Lives, Optimising Resources' report recommended collaboration between rare diseases researchers to ensure that expertise can be shared and limited resources for research into rare conditions can be deployed effectively.

There were also a number of stakeholders who identified in their survey responses a need for public and charitable funding bodies to collaborate with other organisations for research into rare diseases, to help to pool knowledge and resources.

“Rare disease research can compete on a level playing field with common disease research through collaboration and building of strong networks.”

Researcher

Additionally, some responses to the consultation on a UK rare disease plan identified the need for collaboration between different organisations and researchers⁸. Several funding bodies already have schemes which encourage collaboration with industry, patient organisations and internationally in order to encourage research into rare diseases.

The International Rare Cancers Initiative ⁹

The International Rare Cancers Initiative (IRCI) is a collaboration between Cancer Research UK, the UK National Institute for Health Research Cancer Research Network (NCRN), the US National Cancer Institute (NCI), and the European Organisation for Research and Treatment of Cancer (EORTC).

The initiative is designed to support the development of international clinical trials for rare cancers by bringing together clinical communities internationally to identify priorities for research for rare cancers.

The IRCI team hopes that many of these ideas will be developed far enough so that they can be submitted for funding to Cancer Research UK or other research funding bodies.

8. 'Consultation on the UK plan for Rare Diseases: A summary of consultation responses' Department of Health, November 2012

9. <http://www.cancerresearchuk.org/science/research/how-we-deliver-our-research/others/by-programme/international-rare-cancers-initiative>

The Wellcome Trust Pathfinder Awards¹⁰

The Wellcome Trust has been running Pathfinder Awards since April 2012 to provide funding for pilot projects that have significant potential to help develop innovative new products in rare disease areas. The scheme covers any aspect of product development for neglected or orphan diseases.

The scheme awards funding to academic collaborations or not-for-profit organisations with companies that have specialist knowledge and access to technologies to facilitate the development of the product. The company must provide match funding for the project, with either financial or in kind support in money or resources.

The aim of the scheme, besides encouraging effective partnerships between academic or not-for-profit organisations and companies, is to address to an area of unmet need by developing therapies for rare or orphan diseases.

So far funding has been awarded to two projects in collaboration with pharmaceutical companies Pfizer and Lilly.

Recommendation 3: Methods to foster collaboration in the field of rare disease research should continue to be explored and exploited.

Support for researchers

49% of stakeholders who responded to our survey suggested that the application process for funding for research into rare diseases should be made more flexible. However, 51% of interviewees disagreed, stating that all research proposals should be scrutinised in the same way in order to ensure that funding was fairly spent.

Many of our interviewees, especially researchers, said that they wanted to see more support offered by funding bodies to researchers working in the rare disease field throughout research design and the application process as they felt little support is offered currently.

The NIHR's Research Design Service (RDS)¹¹, provides help to those preparing research proposals for submission to peer-reviewed funding competitions for applied health or social care research, free of charge. The service is open to anyone preparing a research proposal, regardless of which funding body they are submitting their proposal to. They have regional offices in England and provide online resources to assist researchers in the following areas:

- Formulating research questions
- Build an appropriate research team
- Involving patients and the public
- Designing a study
- Appropriate methodologies
- Regulatory and ethical issues
- Advice on writing lay summaries
- Identifying the resources required for a successful project.

This is an example of an excellent resource that provides the kind of support rare disease researchers require. However many of the researchers involved in our interviews had not heard of the Research Design Service, or similar initiatives.

10. <http://www.wellcome.ac.uk/Funding/Technology-transfer/Awards/Pathfinder-Awards/index.htm>

11. <http://www.ccf.nihr.ac.uk/Pages/RDSMAP.aspx>

This type of support should be promoted through patient organisations, academic institutions and research networks in the field of rare disease. It is in the best interests of funding bodies to make applicants aware of such support, in order to achieve high standards of applications.

Recommendation 4: Initiatives such as Research Design Service should be better promoted to support researchers in the field of rare diseases.

Peer Review

Peer review is an integral part of assessing all proposals for research, providing independent expert advice on proposals for research. Across all fields of research it is increasingly hard to find peer reviewers, and often hard to find in-depth and good quality peer review¹². One funding body informed RDUK that currently nearly half of all peer review requests are turned down, and that the number of refusals is growing.

Some interviewees, both funding bodies and researchers, reported that this is a particular problem for rare disease research due to the small field of expertise for individual rare conditions.

To address this problem, some funding bodies, such as the Wellcome Trust are considering implementing or trialling peer review colleges across some of their funding schemes. A peer review college is a group of experts who have agreed to peer review a certain number of applications each year for a funding body. This system is currently used by many of the research councils; the Engineering and Physical Sciences Research Council, the Arts and Humanities Research Council and the Economic and Social Research Council. Peer review colleges will provide a guaranteed number of peer reviewers, providing consistent quality and expertise.

73% of those who responded to our survey thought that funding committees should include someone with expertise in rare disease research. However during interviews the necessary broad remit of funding committees was identified as potentially incompatible with ensuring rare disease expertise in the membership. We believe this expertise can be better provided through the peer review system and any peer review colleges should include experts in rare conditions.

Recommendation 5: Peer review of funding applications should include someone with experience in rare disease research.

Our Additional Findings

Patient Registries

A considerable number¹³ of survey respondents across all stakeholder groups agreed that the infrastructure for research into rare diseases is poor. There are significant problems identifying patients to participate in research into rare diseases.

Registries are databases that collect clinical information on particular conditions or groups of conditions. They bring advantages to both research and to patient care; recording prevalence and incidence, the clinical effectiveness of interventions and identifying patients for research¹⁴.

Registries are a key tool in the struggle to understand rare disease and a readily available list of patients makes it easier to recruit volunteers into trials of new therapies. As recognised in the public consultation on a UK rare disease plan: "disease registers allow researchers to understand how the various symptoms develop or abate; how the disease progresses as people with a rare condition get older; and what heralds a fatal outcome"¹⁵. Many of those involved in our research agreed that patient registries were important to research into rare diseases.

12. 'Consultation Response to the Science and Technology Committee: Inquiry on Peer Review' The Wellcome Trust, March 2011

13. Evidence taken from free text responses to the survey

14. 'Patient Registries in the Field of Rare Diseases', Rare Diseases Task Force, April 2009

15. 'Consultation on a UK Plan for Rare Diseases', Department of Health, February 2012

“Disease registries should be a priority.”

Patient

“It is easier to undertake common disease research as the cases are more accessible.”

Researcher

There are currently only 37 national patient registries for rare diseases in the UK¹⁶, out of a total of over 6,000 recognised rare diseases. At present there is no one, obvious source of funding for rare disease patient registries, as they can fulfil many functions. We welcome funding for patient registries from any source, so long as the funding is sustainable and long term and not focused upon a single research question. Traditionally, the public and major charitable funding bodies do not provide funding for such infrastructure; however other funding sources are limited for many rare diseases.

Research and Development Approval

Research projects into rare diseases often have to recruit patients from many different sites, due to the small numbers of people affected by rare diseases. Completing many variable research and development (R&D) approval processes for each of these sites is burdensome, time consuming and often causes delays. Additionally, many interviewees told us that the ethics approval process is also lengthy, complicated and difficult to negotiate for many researchers.

The Integrated Research Application System (IRAS) was created to address some of these problems. However our interviews with researchers revealed that despite efforts to improve and streamline the system of NHS R&D approval is still complex, burdensome, time consuming and expensive.

Often researchers were asked to complete similar forms for each site; the IRAS system brought these into one online system, however researchers indicated to us that there are further forms and information required by R&D offices outside of this system.

For example, for the RAPID (Reliable Accurate Prenatal non-Invasive Diagnosis)¹⁷ project recruitment was delayed by over a year due to the lengthy time taken to receive NHS R&D approval for all recruitment sites.

There are several schemes currently being piloted to further address these issues. For example the Health Research Authority (HRA) is piloting an Ethics Officer scheme¹⁸. The Ethics Officer will provide a number of functions including early assessment of applications and study documentation and the early provision of advice, this aim is to improve favourable outcomes and thus reduce timelines and the administrative burden on researchers and committees.

The HRA are also testing a new system of approval for research that would combine and replace aspects of the current review by NHS R&D and Research Ethics Committees (RECs). The pilot aims to improve timelines, reduce the administrative burden on researchers and committees and improve quality and consistency of ethical review.

Both of these pilot schemes could improve the situation for rare disease researchers, if they take into account the particular problems that they face, such as smaller budgets and the need for multi-site research.

16. 'Patient Registries in the Field of Rare Diseases', Rare Diseases Task Force, April 2009

17. <http://www.rapid.nhs.uk>

18. <http://www.hra.nhs.uk/hra-news-and-announcements/hra-ethics-officer-pilot>

Annex 1: Interview Questions for Representatives of Funding Bodies

The interviews with representatives from funding bodies were guided by the following questions:

- What is the current and potential role of your organisation in funding medical research into rare diseases?
- How does the funding of medical research into rare diseases fit into the policy priorities and strategic direction of your organisation?
- Are there any particular areas of work e.g. specific policies or strategies which are relevant to rare diseases?
- Is funding of rare disease research at all monitored by your organisation and is it possible to find out (or reasonably estimate) how much money your organisation gives to funding medical research into rare diseases?
- How do you assess the quality of an application?
- What is the role of the panel/ peer review reviewers? Do these panels contain rare disease experts?
- Are there any specific issues/ concerns that your organisation has with applications for funding for medical research into rare diseases?
- What is your organisation's policy on giving feedback to research applicants and why?
- Do you think rare disease research would be aided by disease registries for rare diseases? How do you think these registries could be funded?
- What do you think could encourage/facilitate collaboration between researchers? And does your organisation encourage collaboration with between researchers?
- Does your organisation collaborate with other researchers?
- Do you think that specific funding streams for rare diseases would improve rare disease research?
- There is a perception amongst our members that not enough funding is dedicated to research into rare diseases. Do you think this perception is accurate?
- Have you ever come across delays in research into rare diseases' because of complicated research and development approval processes? Are there obvious ways in which you feel the current approval process could potentially be streamlined for rare diseases?
- Are you aware of any best practice examples of rare disease research that have used innovative ways to overcome the problems that rare disease research presents?

Annex 2: Interview Questions for Researchers

The interviews with researchers were guided by the following questions:

- Could you please briefly explain the research proposal(s) you submitted?/Type of research you conduct(ed)?
- Which funding bodies did you submit your application(s) to?
- Why do you think that your application was unsuccessful? OR
- How much funding were you awarded?
- What kind of support did you receive for your application?
- Is there enough support available for rare disease researchers?
- How could funders provide more support to applications?
- What were the criteria for assessment?
- How flexible was the application process?
- Did you receive feedback on your application?
- Was this feedback useful/constructive?
- What do you think UK public/major funding bodies could do to better support rare disease research?

Annex 3: Interviewees

RDUK interviewed the following people for this report:

- Dr Duncan Batty, AKU Society
- Dr Stephen Bloch, UCL Division of Psychology and Language Sciences
- Dr Gillian Borthwick, Collaborative Group for Genetics in Healthcare
- David Dennis, IiH UK
- Dr Audrey Duncanson, The Wellcome Trust
- Dr Tom Foulkes, Medical Research Council
- Dr, Alyson Fox, Wellcome Trust
- Dr Bethan Hughes, The Wellcome Trust
- Kate Law, Cancer Research UK
- Dr Steve Laval, Newcastle University
- Dr Stephen Lynn, TREAT-NMD
- Dr Joe McNamara, Medical Research Council
- Dr Jonathon Sandy, School of Oral and Dental Sciences, University of Bristol
- Dr Tony Soteriou, Department of Health
- Phil Taverner, University of Southampton,
- Mark Turner, Department of Neonatology, Liverpool Women's Hospital
- Dr Susan Walsh, CGD Society
- Dr John Williams, The Wellcome Trust
- Professor Willem H Ouwehand, University of Cambridge and Wellcome Trust Sanger Institute

Annex 4: Survey questions

The following questions were asked in the survey of rare disease stakeholders:

- How do you feel the UK's public and major charitable funding bodies support rare disease research?
- How do you feel the Medical Research Council (MRC) supports rare disease research?
- How do you feel the Wellcome Trust supports rare disease research?
- How do you feel Cancer Research UK supports rare disease research?
- How do you feel other charitable funders (such as British Heart Foundation) support rare disease research?
- Do you think that public and major charitable funding bodies make enough funding available for rare disease research?
- Do you think that public and major charitable funding bodies do enough to make people aware of funding opportunities for rare disease research?
- Do you think there is a level playing field between applications for funding research into rare diseases and research into more common diseases?
- What do you think UK public and major charitable funding bodies could do to better support rare disease research? Please select all that apply:
 - Identify rare disease research as a priority area
 - Improved promotion of the importance of rare disease research
 - Create specific funding streams for rare diseases
 - More suitable/flexible application forms
 - More support for people when making rare disease research applications
 - Supporting the development and funding of research networks
 - Sustainable funding for disease registries
 - Inclusion of someone with expertise in rare disease research on funding committees
 - Nothing
- Do you have any other comments on public funding of rare disease research?

Annex 5: Funding for research into rare diseases

Cancer Research UK reported spending £29.4m on rare disease research between 2011 and 2012, their total for all medical research over the same period was £300 million.

The Medical Research Council reported spending 10% of their translational funding programme on rare diseases, of a total research spent across all programmes of £759.4 million in 2011/2012

The National Institute of Health Research reported spending £6, 964, 785 on research into rare diseases between 2006 -2012, their total medical research spent between 2009 to 2010 was £919.5 million.

The Wellcome Trust reported spending £11.7m on research into rare diseases between 2009 and 2012, out of an average medical research spend of £600 million a year.



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