

RARE DISEASE | UK

Rare Disease Care Coordination: Delivering Value, Improving Services



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



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Particular thanks also to Laura Gilbert for compiling this report and Brenda Kent for interviewing and producing the case studies.

The Rare Disease UK team

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Chair's Foreword

If any of us were in a position to need complex, long-term healthcare, most of us in the UK would probably – and quite reasonably – expect there to be communication and coordination between the professionals looking after us. Yet that is not the reality for the majority of patients and families living with rare diseases.

At Rare Disease UK (RDUK) we hear all too often that patients and families have nobody to coordinate the multiple elements of care and treatment that their condition necessitates. This can mean that they do not receive the information and support that they need, that they have to tell their story over and over to health professionals, or that they feel lost in the healthcare system.

This report sets out how having named care coordinators in post would help to address the unacceptable situation of the majority of patients and families with rare diseases having to battle their way through fragmented care and treatment, through no fault of their own.

As well as drawing on RDUK's own research into the experience of patients and families living with rare diseases, the report outlines the impact of professionals fulfilling the care coordinator role for a number of conditions.

The evidence highlighted in this report shows that having access to a named care coordinator helps to ensure that patients and families receive the well-coordinated care that they should be able to expect. In these straitened times, it would also offer the NHS potential cost savings.

Enabling access to named care coordinators for people with rare diseases makes practical and economic sense. It makes sense for commissioners and providers, who need to deliver quality care and value for money. And it makes sense for the many patients and families throughout the UK who should not have to struggle to navigate the care they need to live with their condition.

It is time for action to be taken so that care for people with rare diseases can be provided for in a joined-up and cost-effective way.



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



Introduction

A rare disease is defined by the European Union as one that affects fewer than five in 10,000 of the general population.¹ One in 17 people, or 7% of the population, will be affected by a rare disease at some point in their lives, which equates to approximately 3.5 million people in the UK.

There are over 6,000 recognised rare diseases, including rare cancers such as childhood cancers, and some other well known conditions such as cystic fibrosis and Huntington's disease.

Often rare diseases are chronic and life-threatening, and have an enormous impact on the lives of patients and their families. Most rare diseases affect multiple parts of the body – such conditions are known as 'multi-system'. Consequently, people with these conditions require complex care from a range of different health professionals.

However, many people with rare diseases tell Rare Disease UK (RDUK) that the care and treatment that they receive is poorly coordinated, which has a detrimental impact on their care and the lives of their families.

RDUK believes that every patient with a rare disease should therefore have access to a named care coordinator.

A care coordinator is a trained professional who is responsible for ensuring that a care plan is in place and carried out. They can greatly assist in ensuring coordination and continuity of care, so that care is provided as smoothly as possible for the patients they support.

Care coordinators are also available to support the practical and emotional needs of patients, families or carers throughout the progression of the condition. They may also provide vital education to other professionals, to enable appropriate care and support to be provided.

Care coordinators may also be known by other job titles, such as care advisors or key workers, or a specialist nurse may fulfil the role. In a UK-wide survey of the experiences of patients and families with rare diseases conducted by RDUK, one fifth (21%) of respondents said that the role of care coordinator should be fulfilled by a specialist nurse.²

However, the lack of an overwhelming preference suggests that as long as there is someone available to centrally coordinate care, patients and families do not mind who it is, as long as the professional has sufficient knowledge of the condition and the situation.

What is crucial is that a sufficient number of care coordinator posts are created, in whatever capacity works best to meet the needs of patients and families affected by rare diseases.

1. European Commission. http://ec.europa.eu/health/rare_diseases/policy/index_en.htm (accessed 9 January 2013)

2. 'Experiences of Rare Diseases: An Insight from Patients and Families', Rare Disease UK, December 2010

Executive Summary

Most rare diseases affect multiple parts of the body, which means that a range of different health professionals need to be involved in providing care and treatment. To ensure that the care required by people with rare diseases is provided as smoothly as possible, coordinated care is essential, as recognised by the UK Health Departments.³

The very nature of rare diseases also means that many people have not heard of their condition when they are diagnosed, and nor have the health professionals they meet. Without someone to coordinate care, it is therefore very difficult for people with rare diseases to manage on their own.

However, only 13% of patients with rare diseases have access to someone to fulfil the care coordinator role. This figure compares, for example, to 87% of people with cancer who are given the name of a clinical nurse specialist⁴, whose role includes coordination. Furthermore, where care coordinator posts are in place for people with rare diseases, resources are often considerably over-stretched.

Consequently, poorly coordinated care is a major issue for the majority of patients and families affected by rare disorders.

Many people with rare diseases tell RDUK that not having anyone to coordinate their care means that they do not receive adequate information and support, or that they feel lost in the healthcare system. In the worst case scenario, a lack of care coordinator support can result in serious mismanagement of care leading to increased hospital admissions.

Providing named care coordinators for people with rare diseases would help to ensure that patients and families receive joined-up care.

RDUK has found strong evidence that care coordinators also represent good value for money for service providers. Their work can lead to significant cost savings, for example by:

- saving consultants' and GPs' time;
- helping to prevent unplanned hospital admissions;
- reducing the length of hospital stays.

Evidence also indicates that having a professional in post who can fulfil the care coordinator role helps to improve quality of care and patients' experience of care, for example by:

- helping patients receive timely access to the specialist knowledge and care they require;
- meeting patients' information needs;
- providing emotional and practical support to patients and their families;
- providing a continuing point of contact.

By enabling patients to have a positive experience of quality care, care coordinators can also help to deliver improvements in care that governments and health services are working towards in all parts of the UK.

Simply put, having named care coordinators in post is an effective – and cost-effective – way of making a great deal of difference to patients and families who deserve better.

3. 'Consultation on the United Kingdom Plan for Rare Diseases', UK Health Departments, February 2012

4. 'Cancer Patient Experience Survey 2011/12 – National Report', Department of Health, August 2012

Why People with Rare Diseases Need Named Care Coordinators

Most rare diseases affect multiple parts of the body, meaning that many different professionals are involved in care and treatment. It is therefore essential to have good coordination and communication between these professionals. However, care for people with rare diseases is currently poorly coordinated and there is a lack of information and support for patients and families. Providing named care coordinators for people with rare diseases would ensure that care is joined up, and that patients and families have access to the support they need to live with their condition.

Most rare diseases affect multiple parts of the body, meaning that many different professionals are involved in care and treatment.

In the 2012 consultation on the UK Plan for Rare Diseases, the UK Health Departments explained how many rare diseases affect multiple parts of the body, necessitating care and treatment from a range of specialists:

'Many rare diseases, particularly those of genetic origin, affect fundamental biological processes in every cell in the body – for example heart, liver, kidney – may be affected, demanding a range of specialist expertise. (...) Some cases may require expertise in a number of disciplines. For example the service caring for people with Biedl Bardet syndrome needs to call on diabetologists, ophthalmologists, nephrologists and all associated diagnostic and therapy services. Similarly, treating Alström syndrome will need to call on cardiologists and transplant services. A different condition such as cystic fibrosis might require a different mix of specialist expertise.' ⁵

It is essential to have good coordination and communication between professionals providing care and treatment for people with rare diseases.

The involvement of many different professionals in providing care and treatment for people with rare diseases means that good coordination and communication between them is crucial, as recognised by the UK Health Departments:

'With so many specialists, and so many hospital departments seeing the same patient to offer their particular expertise, co-ordinated care is essential.' ⁶

5. 'Consultation on the United Kingdom Plan for Rare Diseases', UK Health Departments, February 2012

6. 'Consultation on the United Kingdom Plan for Rare Diseases', UK Health Departments, February 2012

There was 'overwhelming agreement on the need for good coordination of care for people with rare disease' from respondents to the UK Plan for Rare Diseases consultation, with named care coordinators receiving strong support as a solution. 85% of respondents agreed that commissioners of care for people with rare diseases should assess options for improved care coordination, including named care coordinators.⁷

Two major groups of charities also backed care coordinators as a solution in response to the consultation. National Voices, the coalition of health and social care charities in England, recommended that 'the plan includes a commitment to provide a single, trusted and easily contactable point of liaison, or care coordinator, for all people with rare diseases'.⁸ Genetic Alliance UK, the national alliance of over 150 patient organisations supporting all those affected by genetic conditions, took the same view.⁹

"My care is covered by a minimum of 13 people at eight different centres, but the coordinator 'builds a wall' around the patient so that they are not bothered by all the phone calls, emails and bureaucracy in general. The coordinator is in touch with all people concerned with my care. The only appointments I make are with my GP. The amount of stress and worry that this removes from your life cannot be measured."

Patient living with motor neurone disease

The benefits of having a named professional to coordinate care were also recognised over a decade ago in Department of Health guidance on the rare condition Creutzfeld-Jakob Disease (CJD). Like some other rare diseases, CJD is characterised by a combination of symptoms that progress rapidly. The guidance notes that 'Patients with terminal or degenerative conditions benefit from coordination of care and the identification of a key worker.'¹⁰ The role of the key worker for people with CJD is outlined as including coordination of professionals providing care, providing information about the condition as well as emotional support to families and carers. This is precisely the type of support that is also crucial for families with other rare diseases.

In addition, the National CJD Care Team in Edinburgh has two National Care Co-ordinators, who are funded by the Department of Health in England. The Care Co-ordinators, who are senior nurses, work with patients, carers, professionals and organisations across the UK involved in the care of people with CJD, to provide advice, information and support.¹¹

Care for people with rare diseases is currently poorly coordinated and there is a lack of information and support for patients and families.

Poor coordination of care

In RDUK's experience, fragmented, poorly coordinated care is a major issue for the majority of patients affected by rare diseases. Patients can find themselves having to attend hospital – sometimes a number of different hospitals – on different days to see different specialists.

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7. 'Consultation on the United Kingdom Plan for Rare Diseases – Summary of Consultation Responses', UK Health Departments, November 2012
 8. 'Rare disease consultation: National Voices responds'. www.nationalvoices.org.uk/rare-disease-consultation-national-voices-responds (accessed 5 January 2013)
 9. Genetic Alliance UK. www.geneticalliance.org.uk/docs/policy/genetic-alliance-uk-rare-disease-plan-response.pdf (accessed 14 January 2013)
 10. 'Creutzfeld-Jakob Disease: Guidance for Healthcare Workers', Department of Health, 2000. Similarly, 'Creutzfeld-Jakob Disease: Guidance for Health and Social Care Workers', published in 2000 by the Department of Health, Social Services and Public Safety in Northern Ireland, states that: 'Patients with life shortening or degenerative conditions benefit from coordination of care and the identification of a key worker.'
 11. National CJD Research & Surveillance Unit and Care Team information leaflet. www.cjd.ed.ac.uk/documents/leaflet.pdf (accessed 4 January 2013)

The effect on the lives of patients and their families, and their ability to hold paid employment, is extremely detrimental, and is often aggravated by poor communication between the different specialists and other professionals involved in their care. RDUK believes that one of the solutions is for each patient with a rare disease to be designated with a named care coordinator.

However, in a UK-wide survey of the experiences of patients and families with rare diseases conducted by RDUK, only 13% of respondents told us that the person with a rare disease had a designated care coordinator or care advisor. In comparison, recent figures indicate that 62% of people newly diagnosed with Parkinson's receive contact information for a Parkinson's disease nurse¹² and 87% of people with cancer are given the name of the clinical nurse specialist (CNS) in charge of their care¹³.

Not having a care coordinator can aggravate a number of issues for patients and their families. Some of the problems experienced by respondents to RDUK's survey included:

- each professional looking at specific elements of the condition, but no one being concerned with the condition as a whole.
- patients or families having to repeatedly tell their story to all professionals involved in their care.
- feelings of being lost in the healthcare system.
- patients and families not knowing who to go to with queries on their condition.
- a lack of continuity in those involved in the care of the patient.

Care coordinators can help to alleviate these issues.

Insufficient information and support

As evidenced later in this report, where access to care coordinators is available, they are able to address the information and support needs of their patients. It is vital for patients and families with rare diseases to have someone to assist them with their queries as their disease progresses, to offer them support for their practical, social and emotional needs, and to direct them to appropriate services.

However, in response to RDUK's UK-wide survey on patients' and families' experiences of living with rare diseases¹⁴, over half (52%) of the respondents felt they had not been given enough information on their condition following diagnosis.

Our survey also found a lack of support for rare disease patients with their health and social care issues. Over one third (37%) of patients do not have someone they can approach with questions on their condition. And few rare disease patients receive sufficient support to meet their practical and emotional needs:

- only one third (33%) feel that they receive sufficient support with their social needs;
- only 29% feel they receive sufficient psychological support;
- less than one quarter (24%) feel they receive adequate support with financial concerns.

Problems with medical, psychological, financial, social and other issues are frequently experienced by patients at transition periods, notably in the transition from paediatric to adult services, where almost 30% of respondents to our UK-wide survey¹⁵ reported having experienced problems. This figure may actually be higher as a significant number of respondents replied that they 'didn't know' whether problems had been experienced. Furthermore, RDUK's 2013 patient surveys in Scotland and Wales indicate higher proportions of people experiencing problems¹⁶, so financial constraints on health services may be exacerbating transition-related issues.

Having access to a care coordinator would ensure continuity and communication between all necessary people at key transition periods.

12. 'National Parkinson's Audit Report 2011 Summary', Parkinson's UK, May 2012. This report notes that the previous year's audit found that 75% of newly diagnosed patients received contact information for a Parkinson's nurse.

13. 'Cancer Patient Experience Survey 2011/12 – National Report', Department of Health, August 2012

14. 'Experiences of Rare Diseases: An Insight from Patients and Families', Rare Disease UK, December 2010

15. 'Experiences of Rare Diseases: An Insight from Patients and Families', Rare Disease UK, December 2010

16. <http://www.raredisease.org.uk/documents-to-download.htm>

Living with Motor Neurone Disease (MND)

The Motor Neurone Disease Association explains that MND 'is a progressive disease that attacks the motor neurones, or nerves, in the brain and spinal cord. This means messages gradually stop reaching muscles, which leads to weakness and wasting.'¹⁷ Currently there is no cure for MND.

17. www.mndassociation.org/what-is-mnd/About-MND (accessed 16 January 2013)

Arthur's Story

You go in and sit in front of the consultant who confirms you have MND.

"Hey, you must have the wrong fellow. I eat a healthy diet, keep active with swimming, go to keep fit, play outdoor and indoor bowls".

"Sorry. It makes no difference".

What follows is shock, fear, depression - and then you go into denial, not wanting to tell anyone.

I am now in my third year. What has changed in my life? Everything.

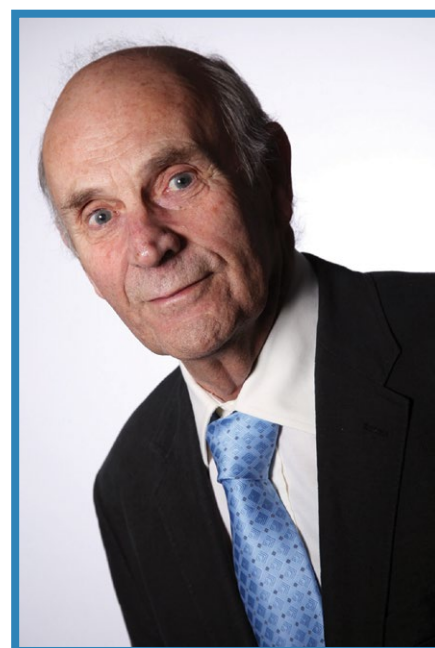
I now have a walking and shooting stick¹⁸, a stair lift, disabled badge and a variety of medications. I can no longer fly to Canada to see my daughter and granddaughters. All my sporting activities are things of the past. On the personal side buttons, zips, shoelaces are things that my wife

has to do for me. All this because of a major problem with shortage of breath and slow loss of body strength.

When I saw the consultant on that fateful day in November 2011, he told me I could expect a call from an MND nurse who would be there to help as and when needed. Not only did I get a call but it was from a coordinator specialising in all progressive neurological conditions.

From there on in things changed. My wife and I felt for the first time that we were not alone. Here was someone very knowledgeable of my condition, and also, and just as important, someone who knew who to ring and when. MND is with you 24/7. There are enough problems coping with the physical changes in your life causing emotional turmoil not previously experienced, without worrying about arranging everything necessary for your care. This is where a coordinator becomes a big part of your life. Also, because of her knowledge, she can arrange appointments for me to see about certain problems without having to go to the GP or consultant.

My care is covered by a minimum of 13 people at eight different centres, but the coordinator "builds a wall" around the patient so that they are not bothered by all the phone calls, emails and bureaucracy in general. The coordinator is in touch with all people concerned with my care. The only appointments I make are with my GP. The amount of stress and worry that this removes from your life cannot be measured. Not only do I get direct support from the coordinator with phone calls and emails and a rare visit to lift my spirits, but she also always



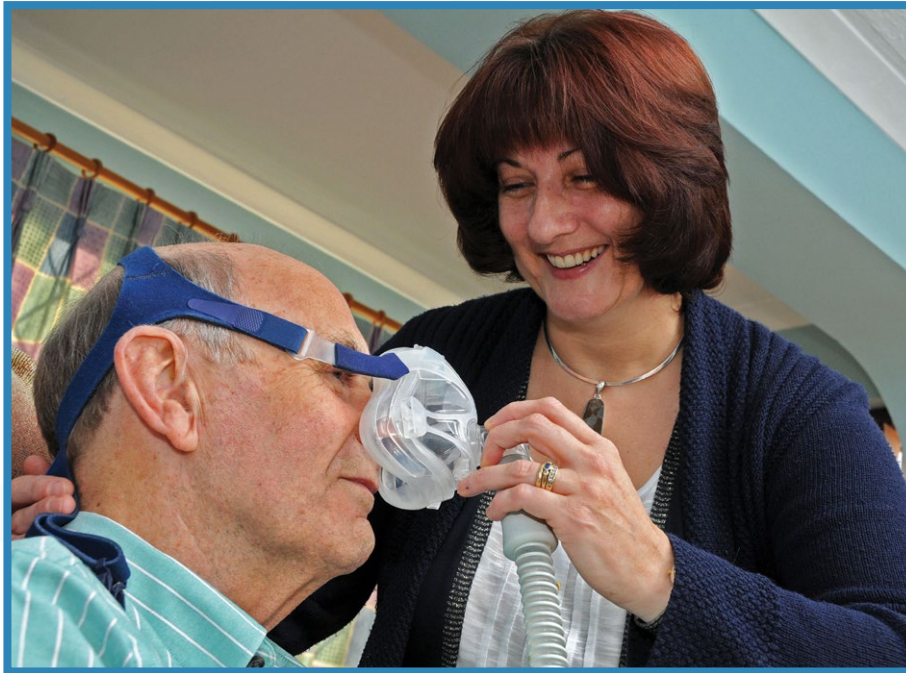
remembers that my wife is in this with me and she has a chat when she can.

Whoever thought of the idea of the post of coordinator deserves a medal. Anyone who thinks this is not a good idea and would want to do away with the position does not understand the first thing about patient care. My wife and I cannot think of life without a coordinator. Just talking to one person takes so much worry out of our lives and leaves us more time to try and enjoy what is left of our lives.

18. A walking stick with a handle that unfolds to form a seat and a sharpened end that can be stuck firmly in the ground

A Care Coordinator's Perspective

Elizabeth Garrod – Care Coordinator with the My Needs Now Project



them and that their developing multidisciplinary team will also use me in the same way. They are unlikely to see me again as my active involvement is usually 'third party' for complex problem solving only and review of cases when I chair the monthly neuro-palliative multidisciplinary meetings.

This simplifies things for both patients and professionals, who benefit from a coordinated service with regular communication and everybody understanding everybody else's role far more clearly. This in turn empowers patients, allowing them control to make informed choices about their care and has increased knowledge of and respect for other professional input within the multidisciplinary team.

Early results from the implementation of the pathway have demonstrated improved access to timely, responsive care from a range of service providers, a reduced rate of emergency admissions to hospital with more patients dying in their place of choice: at home. It has also resulted in an enhanced quality of life for patients and carers.

My background has been palliative care for over twenty years and in September 2012, I became the coordinator for the 'My Needs Now' project, following the successful award of £50,000 from the East of England Transforming Community Services Competition. This successful application was made by statutory services and the third sector jointly identifying the need for the development of a care pathway for rare and/or rapidly progressing neurological conditions.

This is a truly transformational project for patients with rare, progressive neuromuscular conditions who often have

specialist nurses but do not normally have anybody actively working exclusively as I do, in a coordinating capacity.

When someone is referred to the service, I assess how urgent their situation is from the information I have been given before I place a call to determine their needs. Referrals can be made and outcomes achieved without the need for face to face contact, but I will often visit to perform a holistic assessment. I explain that I will act as an expert resource and conduit through which they can access the support and care from all the various services and agencies available to

Delivering Value for Money

Providing named care coordinators for people with rare diseases would require some allocation of resource. However, this would be offset in the short/medium term by increasing efficiency in the way services are utilised, and in the long term by improved patient outcomes.

Care coordinators represent significant value for money; they can save the NHS money across many different areas. The advice, support and information provided by care coordinators for various conditions, both rare and common, have been shown to deliver cost benefits through saving consultant time, preventing hospital admissions and enabling better management of conditions, for example.

The examples outlined below demonstrate how professionals in a care coordinator role deliver value for money in the case of three types of rare condition: rapidly progressing neurological conditions, neuromuscular conditions and sickle cell disorders.

Evidence of cost savings is also provided for care coordinator roles for a number of more common conditions which nonetheless have parallels with rare diseases (e.g. requiring complex care from a range of professionals) and for which support is provided that is also vital for people with rare diseases (e.g. emotional support). It might reasonably be suggested that the potential for cost savings is even greater in the case of rare diseases, given the wide range of professionals involved in rare disease care and the high levels of care needed.

Coordinator for rare and rapidly progressing neurological conditions

The 'My Needs Now' project funded by the East of England Transforming Community Services Competition in Hertfordshire is an excellent example of how the provision of a care coordinator can vastly improve patients' lives while also providing significant cost savings for the NHS.

The project has put in place a coordinated approach, led by a dedicated coordinator, to care for people with rare and rapidly progressing neurological conditions, including motor neurone disease and Huntington's disease. Like many other rare diseases, the nature of these debilitating conditions necessitates complex care and continuing support for patients' and families' changing needs.

The aims of the project are to provide patients with a single point of access to services and support, to simplify the care pathway, empower them to make informed choices, and ensure that service providers are informed about these conditions. The coordinator acts as a resource for professionals involved in the care of the patient, as well as for patients and families.¹⁹

Patients who have had access to the project were 98% less likely to be admitted to hospital, equating to **estimated potential savings of £220,000** over 12 months for the NHS. The team running the project has also noted that an integrated care pathway is seen to reduce the number of hospital admissions and deaths by at least 47%, potentially **saving more than £54,400** per year, with maximum acute services savings of £524,000 per year.²⁰ In addition, evidence from the project indicates that having the coordinator in post has freed up neurology consultants' time²¹, which could enable further cost savings.

19. 'My Needs Now Project – The development of coordinated approach to service provision for people with rare and rapidly progressing neurological conditions living in Hertfordshire', Garrood, E, May 2012

20. 'My Needs Now – Rare and Complex Neurological Conditions Service – update', Garrood, E, November 2012

21. 'My Needs Now Project – The development of coordinated approach to service provision for people with rare and rapidly progressing neurological conditions living in Hertfordshire', Garrood, E, May 2012

Neuromuscular care advisors

As the key worker for patients, neuromuscular care advisors have an important role in coordinating the specialist care needed by people with neuromuscular conditions, which, like many other rare conditions, are diseases. There are 31 neuromuscular care advisor posts across the UK, with full NHS funding.²² However, the Muscular Dystrophy Campaign is calling for the number of posts to be increased to 70²³, in order to meet patients' needs.

The Muscular Dystrophy Campaign has found that neuromuscular care advisors can save the NHS money, 'for example by saving consultants' time, reducing GPs' time, by signposting patients to local services and liaising with other service providers.'²⁴ One consultant in Bristol estimates that having a neuromuscular care advisor in post **saved over 80 hours of consultant time per patient** with Duchenne muscular dystrophy.²⁵ Based on the calculation of one hour of consultant patient-related time at £169²⁶, 80 hours saved equates to **over £13,500 in savings per patient**.

Neuromuscular care advisors can also help to prevent emergency hospital admissions, offering the potential for cost savings, as well as a better experience for patients. The cost of unplanned emergency admissions are estimated to be as high as £68,507,316 in England, £6,829,731 in Scotland, £3,917,052 in Wales and £2,273,534 in Northern Ireland.²⁷ An NHS audit indicates that over a third of neuromuscular hospital admissions are avoidable, and that monitoring of patients and access to services between clinic appointments 'should be strengthened. This could be coordinated in a more formal process by the service, for example by the clinical nurse specialist'²⁸; a neuromuscular care advisor could also undertake this type of coordination.

"I often assist individuals in referral to a specialist centre where their condition can be managed by a multi-disciplinary team. This means that they can be seen on the same day by different medical staff and this saves time and resources. Furthermore, as they are seen by specialists, there are fewer emergency admissions as their condition is managed better and consequently they have a better quality of life."

Pammy Malhotra, Neuromuscular Care Pathway Coordinator and Patient Advocate for the Muscular Dystrophy Campaign

Sickle Cell and Thalassaemia Specialist Nurses

Sickle cell and thalassaemia specialist nurses, whose posts are NHS-funded, work both in the community and acute trusts, providing support for patients with rare, sickle cell disorders and thalassaemia. The nurses assist patients and families through care planning, promoting self-management and brokering care with a range of professionals, for example.

A study in 2012 of the contribution of sickle cell and thalassaemia specialist nurses in England²⁹ indicated that the nurses freed up consultant time by running outpatient clinics, and that their work may also result in

22. Muscular Dystrophy Campaign. www.muscular-dystrophy.org/how_we_help_you/care_and_support/care_advisors (accessed 7 January 2013)

23. 'Annual review 2011/12', Muscular Dystrophy Campaign

24. 'Invest to Save: Improving services and reducing costs', Muscular Dystrophy Campaign, May 2011

25. 'Invest to Save: Improving services and reducing costs', Muscular Dystrophy Campaign, May 2011

26. 'Unit Costs of Health and Social Care 2010', Personal Social Services Research Unit, 2010

27. 'Invest to Save: Improving services and reducing costs', Muscular Dystrophy Campaign, May 2011

28. 'Unplanned admissions of neuromuscular patients – a collaborative audit', NHS Audit, Information and Analysis Unit, June 2012

29. 'Understanding the contribution of sickle cell and thalassaemia specialist nurses', Prof Elizabeth Anionwu & Dr Alison Leary, supported by Roald Dahl's Marvellous Children's Charity, July 2012

unscheduled emergency hospital admission being avoided, for example through supported self-management strategies. Such activity may generate potential cost savings (although it should be noted that the nurses in the study recorded a mean unpaid overtime of three hours per week).

Clinical nurse specialists (CNSs) in cancer

As is the case with rare diseases, the care needs of people with cancer can be highly complex, necessitating contact with a number of different health professionals. Between 30% and 50% of cancer cases could in fact be classed as 'rarer'.³⁰

The support provided by CNSs in cancer – often known as 'Macmillan nurses' where Macmillan Cancer Support has provided pump-prime funding for CNS posts – includes coordinating care, providing information and offering emotional support for people with cancer and their families/carers.

Macmillan has found that 'CNSs represent good value for money by reducing the number of emergency admissions, the length of hospital stay, the number of follow-up appointments, the number of medical consultations and providing support to enable people to be cared for and die in their place of choice.'³¹

The potential cost savings of having a CNS in post could be significant. A report prepared for the Department of Health estimated the cost of workforce expansion to provide one to one support for all cancer patients in England at £60 million per annum, which would be offset by **expected savings of £89 million** from improvements in quality and coordination of care.³²

Rheumatology nurse specialists (RNSs)

NHS-funded RNSs support people with conditions such as rheumatoid arthritis, which share some similarities with rare diseases, such as the unpredictable nature of the condition and the need for complex care. As well as providing clinical expertise, RNSs support patients by coordinating complex care and providing psychological support, for example.

A study by the Royal College of Nursing on the work of RNSs across the UK shows that RNSs 'represent good value for money, through reducing costs in primary care and saving consultants' time.' Per annum, outpatient work by RNSs is worth £72,128 per nurse, and **saves £175,168 per nurse** by freeing up consultant appointments. In addition, telephone consultations by RNSs reduce the number of GP appointments, **saving £72,588 per nurse**.³³

Parkinson's nurses

Although Parkinson's disease is a more common condition, it has much in common with rare diseases: it is a degenerative condition for which there is currently no cure, the care pathway is complex, and contact is required with a range of different services and professionals. Similarly to rare diseases, the impact of Parkinson's disease on the lives of patients and their carers means that emotional support is also important.

The charity Parkinson's UK provides pump-prime funding for Parkinson's nurses, who work in a variety of settings across the UK, including hospitals, care homes and the community. The nurses provide practical and emotional support for people with Parkinson's disease and their carers, work with their patients to coordinate the range of services and professionals involved in their care, and provide a continuing point of contact.

Parkinson's nurses can save significant time and resources by providing support, guidance and assistance to people with Parkinson's. For example, a Parkinson's UK report on Parkinson's nurses in England shows that **one nurse can save an average of £80,000** in unplanned hospital admissions by supporting patients at home and in the community – which would equate to estimated savings of over £4.8 million per year if

30. Rarer Cancers Foundation. www.rarercancers.org.uk/index.php?option=com_content&view=article&id=1&Itemid=2 (accessed 5 January 2013)

31. 'Cancer Clinical Nurse Specialists: An evidence review', Macmillan Cancer Support, November 2011

32. 'One to one support for cancer patients', Frontier Economics, December 2010

33. 'Clinical nurse specialists: adding value to care – An executive summary', Royal College of Nursing, April 2010

'replicated across the NHS in England, with a further 60 nurse posts'. By reducing the length of hospital stays, a Parkinson's nurse can also **save an average of £147,000**.³⁴

In other parts of the UK, nurses have reduced hospital admissions by 10%.³⁵ In Scotland, for example, a Parkinson's nurse team has **saved around £39,000** over 18 months in avoided hospital admissions, by providing early stage intervention.³⁶

By freeing up consultants' time, a Parkinson's nurse can also save an average of **£43,812** in consultant appointments – a saving of over £2.63 million per year if 'replicated across the NHS in England, with a further 60 community based nurse posts'.³⁷ In Wales, examples of savings include a Parkinson's nurse saving around **£54,000** of outpatient clinic costs in one year.³⁸ In Northern Ireland, it is estimated that a Parkinson's nurse would save **at least £30,000** of consultant appointment costs per year.³⁹

34. 'Protect Parkinson's nurses in England', Parkinson's UK, June 2011

35. 'The contribution of Parkinson's nurses in Northern Ireland: a guide to equitable services', Parkinson's UK, February 2012

36. 'Parkinson's nurses in Scotland: providing effective, safe, person-centred care', Parkinson's UK, April 2012 The Parkinson's nurse team in question is in NHS Ayrshire and Arran, and comprises two nurses.
www.parkinsons.org.uk/local_to_you/find_local_branches/scotland/ayrshire_branch/ayrshire_-_news.aspx (accessed 23 January 2013)

37. 'Protect Parkinson's nurses in England', Parkinson's UK, June 2011

38. 'Parkinson's nurses – affordable, local, accessible and expert care: A Guide for planners in Wales', Parkinson's UK, June 2011

39. 'The contribution of Parkinson's nurses in Northern Ireland: a guide to equitable services', Parkinson's UK, February 2012

Living with Tuberos Sclerosis (TSC)

The Tuberos Sclerosis Association explains that: 'TSC is a complex genetic condition caused by an alteration in a gene. People with TSC have growths in different organs of the body (brain, heart, eyes, skin, kidneys, lungs) and may have epilepsy, learning disabilities, autism spectrum disorder and kidney problems.'⁴⁰

40. www.tuberous-sclerosis.org/ (accessed 16 January 2013)

Christine's Story

Christine Haig – mother and carer of Aileen who has Tuberos Sclerosis Complex

Aileen was born in November 1967 and although she had her first seizure at one year and eight months old, she wasn't diagnosed with Tuberos Sclerosis Complex until just before her tenth birthday. TSC is a complex condition in which tumours can grow on different organs. It can cause absolute mayhem, but how badly affected a person is depends on the organs affected and the size and number of the tumours. In Aileen's case, she is quite severely affected physically and has learning difficulties as well.

When you have a child seriously affected by a rare disease that even some doctors haven't heard of, some of that carefree spirit that every person has dies. All the plans you had for that child die too. The grief that you feel, you can almost taste it. And if you aren't given the information you need, you don't even know what you are facing. I felt like Calamity Jane: I seemed to lurch from one calamity to another. In my case, I was given the diagnosis over the phone at work. I then went to the university bookshop and looked the condition up in a medical encyclopaedia and a medical dictionary. I have no recollection of putting those books down and driving back to work.



condition, who can put doctors in touch with experts able to give them additional information about TSC, and who knows what the various professionals and services should be offering, and who can offer emotional and practical support and advice to enable me to make informed decisions.

You don't want tea, cake and platitudes; you are looking for help with a desperately sick child or adult. You

Being referred to experts in centres of excellence should be automatic for people with rare diseases and would save money because so often you only take up time at the local hospital and get absolutely nowhere. Nobody should have to go through the trauma and frustration of beating your head against a wall in that way.

So how does having a Tuberos Sclerosis Adviser help?

I can't emphasise enough the devastation you can feel when you have got this desperately sick child or adult. If you could run away, you would run away. But to have someone at the end of the phone who knows about this condition, like our Tuberos Sclerosis Adviser, relieves the panic. You've got someone who is very knowledgeable on the physical and psychological aspects of the

want positive help like the help we get from our TSC Adviser and from the experts at the study days she organises on the various problems that can arise. We recently had one on behavioural problems. It was very down-to-earth and we were given really practical advice on how to handle many different scenarios.

On one occasion before we had a TSC Adviser, Aileen was discharged from the local hospital by a doctor who thought her symptoms were psychosomatic. In fact, she had had a bleed from a tumour on her kidney. If we had had a TSC Adviser at the time, this would never have happened. Now, no matter what happens to me and Aileen, I've got a backup: someone who will help us in a tight corner.

A Care Coordinator's Perspective

Lynn Shields – Tuberous Sclerosis Adviser

My post is funded solely by the Tuberous Sclerosis Association (TSA), and involves working with and supporting people in Scotland living with Tuberous Sclerosis Complex (TSC) and their carers. Current estimates place TSC affected births at 1 in 6000, and diagnosis may follow the identification of heart tumours, infantile spasms in babies or may only become apparent later in someone's life.

People often comment that prior to contacting the TSA, they struggle to access information on TSC and experience a lack of understanding and support for both their medical and non-medical needs. They report that their care is badly coordinated at times and that they face difficulties in accessing the services they require to manage this variable and complex condition. Unfortunately uninformed decision making can lead to the mismanagement of TSC, potentially resulting in the deterioration of an individual's health and, on occasion, crisis visits to A&E.

I work with families, statutory service providers and voluntary organisations in an anticipatory way to improve outcomes. I often arrange and attend multi-disciplinary meetings attended by families together with education, health and social work professionals. The aim is to encourage effective collaboration and communication between everyone involved. It's incredibly rewarding to be invited to support families as their needs change, and to help them access appropriate services and information, including medical, research, education and social welfare.

TSC can affect multiple body systems, so often many health professionals need to be involved

in an individual's care and treatment. Fortunately families in Scotland now have access to a TSC Clinic in Glasgow at the Southern General Hospital. I attend the clinic, which is coordinated by Dr S Joss, and meet many families there. It gives them access to multiple specialisms, including genetics, urology and psychiatry. It's vital that there are links between all those involved and the clinic provides a multi-disciplinary approach and coordination of families' care, resulting in efficient use of NHS resources and increased patient/family satisfaction.

I also produce a Scottish newsletter which includes relevant updates on policy and practice in Scotland. In addition, I organise and manage our Scottish Annual Meetings and a variety of parent and carer information and training and awareness events. Feedback from families and professionals has been the catalyst for these events. Families report the benefits include a greater understanding of challenging behaviour, epilepsy and autism, and feeling better equipped to cope with their often daily challenges. Families also commented that they left our last Annual Meeting in Glasgow, where the focus was the latest TSC research, with 'a sense of hope for the future'. Thankfully, I can liaise with the TSA's Board of Professional Advisers, who advise on research projects and other medical and scientific aspects related to TSC.

It is important to remember that people with TSC and their families

have to live with neurocognitive and behavioural challenges every day that often go unrecognised. It is the manifestations which aren't tumours that have the most far-reaching impact. 'Challenging behaviour' is a deceptively upbeat description for the rollercoaster that many TSC families find themselves on. I spend a great deal of time offering supporting, and discussing these issues with families and statutory services.

I have had the pleasure of supporting a number of inspiring individuals in Scotland and recognise that TSC impacts on each individual and family in a unique way. I have also worked in mutually



beneficial partnership with many other voluntary organisations. I believe working together and making the best use of expertise and resources, particularly in the current climate, represents value for money and, most importantly, empowerment for TSC families. I would suggest mechanisms should be in place from Government to ensure sustainable funding for this approach.

Living with Duchenne muscular dystrophy (DMD)

The Muscular Dystrophy Campaign explains that: 'DMD is a neuromuscular condition caused by the lack of a protein called dystrophin. It is a serious condition that causes progressive muscle weakness. About 100 boys with DMD are born in the United Kingdom each year.'⁴¹

41. <http://www.muscular-dystrophy.org> (accessed 30 January 2013)

Rebecca's Story

Rebecca Kinge – mother of Ethan, who has DMD

We had our concerns that Ethan wasn't achieving as well physically as we would have expected from the time he was six months old. Our concerns grew and by the time he was two years old, we had contacted the paediatrician.

Since his diagnosis, Ethan's mobility has improved, which is usual in DMD boys. He now takes steroids to aid his mobility and health. However, he finds it difficult to walk when he is tired. He falls down more frequently than his peers and finds it difficult to get up to standing from a sitting position. He is unable to run, skip or hop, which is very difficult when he is with his peers or taking part in Physical Education at school.

Ethan has one-to-one support at school as he needs help opening doors, dressing himself and moving around the school amongst the other students. His condition affects all parts of his life, but he manages to cope really well with his determined, positive character.

Our care coordinator has been of huge support since my son was diagnosed. All of Ethan's physical, social, intellectual and emotional needs have been addressed with urgency and care.

Diagnosis is one of the worst times for parents of a little boy with DMD and our care coordinator was there every step of the way to advise, support and basically get us all through! Since then she has gone into my son's school to talk to pupils and staff, helped us to get specialist equipment, supported us all through appointments and been a shoulder to cry on when things aren't going so well.

Our care coordinator is invaluable to the whole family. She has become a friend who knows our family and how we tick. She is at the end of a phone whenever and wherever we need her. She is the first port of call when there is a question or a need. She has an extensive knowledge of my son's condition and knows what he requires and when. She has built up a good relationship with my son that will ensure that as he grows up, he knows he can rely on her too, just as I do.

If the care coordinator wasn't there, the anxiety and worry of taking care of my son would be increased. She has helped us to meet other parents in the same situation as ourselves as well as organising information days, which answer any nagging questions or doubts. She has been



there to give support and advice, and to give emotional stability. She has always been happy to assist with any problem that my son has experienced.

Life would be far more difficult without her.

A Care Coordinator's Perspective

Pammy Malhotra - Neuromuscular Care Pathway Co-ordinator & Patient Advocate

I work as a Neuromuscular Care Pathway Coordinator and Patient Advocate for the Muscular Dystrophy Campaign and my post is funded by the NHS. I provide support and advice to patients and their families on the South East Coast, which includes Kent, Surrey and Sussex.

My job involves attending adult and paediatric clinics at London Hospitals and also outreach clinics around the South East Coast. Furthermore, it involves visits to individuals' home, school or place of work to assist and advise them. It also entails signposting patients to relevant specialist centres for their condition so that they receive the best care from a multi-disciplinary team.

It is very satisfying to see how beneficial my involvement is. An important aspect of my role is that I am able to visit individuals and/or families in their home, school or work environment. I feel this is valuable: firstly, to the individual as they are able to access support and advice directly, and secondly, to myself as, I am able to see the individual in an environment in which they are comfortable - and they do not have to travel which is difficult with muscle conditions. It is also obviously important if home adaptations are being looked at.

Individuals have different issues. Therefore it is important to get a

complete picture by meeting them. On numerous occasions I have found that a person is not under a specialist centre for their condition and this in turn has caused them more problems and distress. This as well as other issues have a detrimental effect on their quality of life

It is very unfortunate that there are so many people that have 'got lost in the system': a recent audit of unplanned hospital admissions revealed that a large proportion of patients were unknown to service providers, and are therefore not accessing a care coordinator for their condition.

I definitely think care coordinators represent value for money. I often assist individuals in referral to a specialist centre where their condition can be managed by a multi-disciplinary team. This means that they can be seen on the same day by different medical staff and this saves time and resources. Furthermore, as they are seen by specialists, there are fewer emergency admissions as



their condition is managed better and consequently they have a better quality of life.

I believe that a care coordinator is important to patients with muscular dystrophy as they then have one person they can turn to for support and advice. A lot of my patients or families have said that it is difficult enough sometimes managing the condition but having to do it all on their own would be impossible.

Improving Patient Experience and Quality of Care

Care coordinators can help their patients to have a positive experience of the care they receive, by ensuring the effective coordination of services, providing a vital source of information and support, and assisting effective self-management. They also provide a point of contact for questions and advice about patients' care, which reduces the sense of isolation that people affected by rare diseases often feel.

“Traditionally it was not uncommon for patients to feel angry and frustrated with the care they received due to the lack of understanding and knowledge around their condition. The presence of a nurse specialist has allowed patients to access a person who understands their condition to discuss their care and answer any questions or concerns they have and thus allowing patients to take an active role in their healthcare.”

Steven Wise, Renal Metabolic Nurse Specialist

The positive impact that care coordinators can have not only on patients' experience of care, but also on quality of care complements a number of policy initiatives and drivers to improve care in all parts of the UK. For example:

- In **England**, the Government's mandate to the NHS Commissioning Board includes a call to focus on more joined-up care for people with long-term conditions: 'We want to see improvements in the way that care: is coordinated around the needs, conveniences and choices of patients, their carers and families – rather than the interests of organisations that provide care; centres on the person as a whole, rather than specific conditions; ensures people experience smooth transitions between care settings and organisations, including (...) children's and adult services'⁴². Furthermore, current government reforms of the care and support system include a focus on better integration of health and care services, recognising that 'people often feel 'bounced around' and have to fight the system to have the joined-up health, care and support they need'.⁴³ Ensuring access to named care coordinators would help to deliver these improvements for those affected by rare diseases.
- The Healthcare Quality Strategy for NHS **Scotland** sets out six quality ambitions including providing person-centred 'care that is responsive to individual personal preferences, needs and values and assuring that patient values guide all clinical decisions'.⁴⁴ Providing named care coordinators would assist NHS Scotland in meeting this ambition.

42. 'The Mandate: A mandate from the Government to the NHS Commissioning Board: April 2013 to March 2015', Department of Health, November 2012

43. 'Caring for our future: reforming care and support', Department of Health, July 2012

44. 'The Healthcare Quality Strategy for NHS Scotland', The Scottish Government, May 2010

- In **Wales**, the NHS improvement programme aims to ensure a person-centred focus whereby: ‘Individuals will be encouraged to take responsibility for their own health and management of conditions, supported with clear and consistent information on how to achieve this.’⁴⁵ Support from named care coordinators would greatly assist people with rare diseases to have clear and consistent information to help manage their conditions as they progress.
- A 2012 policy framework for people living with long-term conditions in **Northern Ireland** outlines six principles, one of which concerns improving care and services for managing long-term conditions. This principle includes providing person-centred services and the delivery of co-ordinated treatment, care and services through collaborative working.⁴⁶ The provision of named care coordinators would be fully in line with this principle.

Examples are outlined below of how care coordinators for people with rare neurological, neuromuscular and sickle cell disorders help to provide patients with a positive experience of care and contribute to the quality of care. Examples are also provided of how specialist nurses fulfilling a care coordinator role improve experience of care for people with more common conditions which share similarities with rare diseases.

Coordinator for rare and rapidly progressing neurological conditions

As described previously in this report, the My Needs Now project in Hertfordshire has implemented a coordinator-led approach to care for people with rare and rapidly progressing neurological conditions, including MND and Huntington’s disease. A key driver for setting up the project was to improve patient experience and satisfaction with care by developing a coordinated care pathway, as outlined in the National Service Framework for Long Term Conditions.⁴⁷

Feedback from patients and carers who have accessed the project has been very positive, for example with regard to helping them receive timely access to the specialist knowledge and care that they need. The project has in fact resulted in an increase of more than 100% in patients accessing neurological services compared to the previous twelve months.⁴⁸

The project also supports patients with self-management strategies, both to empower them and reduce their clinical needs. The support needs of carers are assessed and identified, too, with a view to reducing carer breakdown.

The impressive clinical outcomes achieved by the project include patients reporting that their quality of life is enhanced, and 100% of patients who state a preference of being able to die in their preferred place of death.⁴⁹

Sickle Cell and Thalassaemia Specialist Nurses

A study examining the work of Sickle cell and thalassaemia specialist nurses⁵⁰ found that patients, families and carers value the contribution of the specialist nurses, for example, in coordinating and managing care and being the ‘key accessible professional’. Comments from patients and families clearly indicated that the nurses’ work impacts on the quality of care received.

The study also showed that a significant part of the work of the specialist nurses focuses on psychosocial support, and that psychological support provided by the nurses focused on managing patients’ anxiety and meeting their information needs. This type of support is required by many people with other rare diseases as well as their families/carers.

45. ‘Achieving Excellence: The Quality Delivery Plan for the NHS in Wales 2012-2016’, Welsh Government, May 2012

46. ‘Living With Long Term Conditions – A Policy Framework’, Department of Health, Social Services and Public Safety, April 2012

47. ‘My Needs Now Project – The development of coordinated approach to service provision for people with rare and rapidly progressing neurological conditions living in Hertfordshire’, Garrod, E, May 2012

48. ‘My Needs Now – Rare and Complex Neurological Conditions Service – update’, Garrod, E, November 2012

49. ‘My Needs Now – Rare and Complex Neurological Conditions Service – update’, Garrod, E, November 2012

50. ‘Understanding the contribution of sickle cell and thalassaemia specialist nurses’, Prof Elizabeth Anionwu & Dr Alison Leary, supported by Roald Dahl’s Marvellous Children’s Charity, July 2012

Neuromuscular care advisors

Neuromuscular care advisors throughout the UK act as the key worker for people with rare neuromuscular conditions such as MND. The care advisors provide advice and support for patients and families, and they are the main point of coordination between different health professionals involved in providing care.

The Muscular Dystrophy Campaign notes that: 'Care co-ordination can be vital to the quality of service received.'⁵¹ It is also noted that neuromuscular care advisors 'play a vital role in improving patient care by taking responsibility for co-ordinating the specialist care they receive.'⁵²

"The most satisfying part of my job is in making a difference and improving the quality of life for the patient and their family."

Neuromuscular care advisor in the East of England ⁵³

Clinical nurse specialists (CNSs) in cancer

Evidence indicates that CNSs in cancer, who provide emotional and practical support as well as coordinating care, have a very positive impact on patients' experience of care. The 2011/12 Cancer Patient Experience Survey collected responses from nearly 72,000 people in England about their experience of cancer treatment and care. 87% of patients said they had been given the name of the CNS in charge of their care, and patients with a CNS gave more positive responses on almost all survey questions than those without a CNS.⁵⁴

Macmillan Cancer Support notes that the role of CNSs includes providing information and liaising with other professionals 'to improve the cancer care process for patients', and that 'CNSs can help to improve quality of life for people with cancer through assisting with decision making, symptom management and emotional support.'⁵⁵

Like cancer, rare diseases have an enormous impact on the lives of patients and their families, and they can be life-limiting. Having access to someone who can fulfil the care coordination role and provide emotional and practical support is therefore just as important for rare disease patients' experience of care.

Rheumatology nurse specialists (RNSs)

The work of RNSs includes looking after the physical and emotional needs of people with conditions such as rheumatoid arthritis, and evidence suggests that they have a beneficial impact on patient experience and quality of care. For example, analysis conducted by the Royal College of Nursing shows that patient outcomes from RNSs' practice include: alleviation of physical and psychological suffering, assessing and meeting patients' information needs, and providing 'access to a key contact/knowledgeable professional, or brokering rapid access to another professional'.⁵⁶

These types of support are also key for people with rare diseases, which have several characteristics in common with rheumatological conditions, including the need for psychological as well as physical support.

51. 'Invest to Save: Improving services and reducing costs in Scotland', Muscular Dystrophy Campaign, Sept 2011

52. 'Invest to Save: Improving services and reducing costs', Muscular Dystrophy Campaign, May 2011

53. 'Annual review 2011/12', Muscular Dystrophy Campaign

54. 'Cancer Patient Experience Survey 2011/12 – National Report', Department of Health, August 2012

55. 'Cancer Clinical Nurse Specialists: An evidence review', Macmillan Cancer Support, November 2011

56. 'Clinical nurse specialists: adding value to care – An executive summary', Royal College of Nursing, April 2010

Parkinson's nurses

Parkinson's UK has called for more Parkinson's nurse posts to ensure adequate access to support, noting that the nurses are viewed by those with Parkinson's disease as 'the most important person in helping them manage their condition'.⁵⁷

NICE Guidelines on Parkinson's disease underscore the impact that Parkinson's nurses can have on the care their patients receive. The guidelines recommend referral 'to a specialist, which may be a Parkinson's nurse', for services including 'providing a continuing point of contact for support' and providing 'information about clinical and social issues'.⁵⁸

The guidelines also note 'that "people with long term neurological conditions have improved outcomes and a better quality of life when they are able to access prompt and ongoing advice and support from practitioners with dedicated expertise"'.⁵⁹ NICE's view would apply equally to people with rare diseases, many of which share similarities with long-term neurological conditions.

57. 'Protect Parkinson's nurses in England', Parkinson's UK, June 2011

58. 'Parkinson's nurses – affordable, local, accessible and expert care: A Guide for planners in Wales', Parkinson's UK, June 2011

59. 'Parkinson's nurses – affordable, local, accessible and expert care: A Guide for planners in Wales', Parkinson's UK, June 2011

Living with Primary Immune Deficiency (PID)

The UK Primary Immunodeficiency Support Group (UK PIPs) explains that “The term Primary Immune Deficiency (PID) means a disease that has happened because of a fault (usually a genetic fault) in the body’s ability to either produce or maintain a complete immune system. Just as there are many different parts of the immune system, so there are many different types of Primary Immune Deficiency.”⁶⁰

60. <http://www.ukpips.org.uk> (accessed 18th February 2013)

Stella’s Story

After many years of suffering from throat, sinus and chest infections, “unusual” skin infections and a general lethargy when even my friends and family laughingly called me “hypochondriac”, I was finally diagnosed with a PID in 2011, as well as a partial MBL deficiency.

In layman’s terms, this means that my immune system does not recognise certain types of bacteria as being “invaders”, so my body does not respond by making antibodies. Apart from these “invaders” running wild within the system when you get a “bug”, they can cause extensive damage to the internal organs (such as the chest, ear drums etc). For some people, the disease can be fatal.

It was such a relief to be diagnosed. I was always known as the ‘one who is off sick’, or ‘always ill’. When I have to cancel plans due to sickness, even now, some people think it is just an excuse not to do something.

I spent 15 years seeing various GP’s for immune system tests and they all came back as fine. But I knew in my heart that there was something was wrong with me, as I was permanently ill and tired.

My condition is managed by permanent antibiotics which build a “barrier” between my immune system and a bacterial infection (though cannot stop viruses) and nasal steroid sprays. I have also

had vitamin D injections to help boost my immune system.

My PID has a devastating effect on my life. Luckily, I work for myself running an internet business, and the way I manage my illness and time doesn’t impinge on my clients. But in my personal life, I rarely have the energy to socialise or else I have to cancel plans due to illness.

My care coordinator, has been a lifeline for me. I first met him when I was having my initial round of tests at the special immunology clinic at Barts Health NHS Trust, in London. He listened to me, treated me with respect and all my symptoms as ‘real’ – even though nothing had been confirmed at that stage. He was understanding, compassionate and kind (and still is). At that first meeting, he even gave me his email address so I could contact him while waiting for my results – something unheard of when using the NHS or private treatment.

He also suggested resources I could use to find out more about my condition, once diagnosed.

Since then, he has been my “go to guy” if I have any worries or concerns. I can pick up

the phone to him or email him, and he’ll help, whether I need advice, more medications or test results. He stops me feeling isolated and, without him, I’d feel at a loss to be honest.

All the staff at the clinic at Barts Health NHS Trust do a tremendous job, but without one main person who knows you and who you know you can contact whenever you need to, it can be difficult. My care coordinator provides practical and emotional support.



A Care Coordinator's Perspective

John Dempster – Clinical Nurse Specialist for Patients with Primary Immune Deficiencies (PID)

I work as a clinical nurse specialist, caring for patients with primary antibody deficiencies. As part of my role, I educate and train patients to self-administer intravenous and subcutaneous immunoglobulin at home. I also help to coordinate their care, together with immunologists and community services. My post is funded by the NHS trust where I work.

Another important aspect of my role is to act as a specialist resource and I am available on the telephone and via email to patients and health care professionals to give advice on the condition and its treatment as well as assisting in coordinating care.

I feel that my work has a hugely beneficial impact upon the quality of life of patients. This is because part of my role is to help facilitate patient-centred care through the education and training I provide, which helps patients to stay well and healthy.

Another benefit is the home therapy programme that I coordinate, which enables patients to perform treatment regimens at home at a time convenient to them and therefore reduces the need for time off school and work to visit a healthcare facility. Patient feedback questionnaires have

highlighted the value of this as well as the importance of having a clinical nurse specialist involved in their care.

Primary immune deficiencies are rare medical conditions and are generally poorly understood by the wider healthcare community. It is essential, therefore, to have a care coordinator who is experienced in these conditions and who can advocate on behalf of patients.

I think that care coordinators represent value for money as they are usually the first point of contact for patients if any problems arise. As a clinical nurse specialist, I am able to assess patients' problems via the telephone and deal with them appropriately. This often involves providing advice regarding health, liaising with other healthcare professionals in the community and advising on antibiotic treatment – all of which can prevent a deterioration of a patient's health and an admission to hospital.



Having a care coordinator for patients with PID can help to assist patients' transition from hospital to homecare, result in quicker access to treatments and consequently help to provide a more cost-effective and patient-centred model of treatment, thus helping to reduce costs and improve quality of life for patients.

Living with a metabolic condition

Living with nephropathic cystinosis Ami's Story

I was diagnosed with nephropathic cystinosis, a genetic metabolic condition, when I was 22 months old. The condition led to my kidneys failing and it was predicted that I would need a kidney transplant by the age of five. Fortunately, due to the development of cysteamine therapy, which helped to slow down the rate of kidney failure, I didn't need my first transplant until I was 23 when I received a kidney from my mother.

The condition also affects my eyes. I am very light sensitive due to cysteine accumulation in my eyes. This means I have to use eye drops several times a day. If I did not use the eye drops at all, the cysteine accumulation could lead to blindness.

I also developed scoliosis as a teenager, which required surgery. I don't know if this was related to cystinosis or if it was just an unfortunate coincidence. As nephropathic cystinosis is so rare and there is a lack of research, there is still a lot that is not understood about it. I do believe I would have been taller if I did not have cystinosis, as the rest of my immediate family are relatively tall and I am quite small.

Considering the prognosis when I was diagnosed, I am relatively well at the moment. The main impact my condition has on me currently is the need to try and maintain my health by taking a variety of different medications. I also have to attend a variety of different hospital appointments

for check-ups. As the condition progresses it can lead to other health problems which I do not currently have, such as muscle wastage affecting a person's ability to speak, swallow and also their fine motor skills. Although this is something that does not affect me currently, it is a concern of mine, and I worry about having these types of issues as I get older as they would impact on my ability to work and live independently.

Having a care coordinator has enabled me to see someone for my cystinosis who knows what the condition is and how it affects the body. Previously I only had care from renal services even though my condition is not a kidney disease but a metabolic condition which affects every cell in my body. It has also been really helpful to be able to undergo a variety of tests on different areas of my body in order to identify if there were any underlying issues that I wasn't aware of or if my cystinosis had started to impact on other parts of my body. My care coordinator helps me to organise all the different appointments and to schedule the tests over the course of one or two days so that I can have my whole body checked out at once - sort of like an annual 'MOT'.

Fortunately, these tests have all



come back so far with good results that have helped to reassure me that I do not need to worry about these other areas. If it was not for being able to have these tests, it is likely that I would be unaware of any problems in these areas developing until they became much worse and I started to develop symptoms. They would then most likely be much more difficult to treat.

It is good to have a contact who can provide me with information about my condition, rather than what I have been used to previously: which was usually me having to tell doctors and other health professionals about my condition.

A Care Coordinator's Perspective

Steven Wise Renal Metabolic Disease Nurse Specialist

Traditionally care for patients with rare renal diseases has been patchy, fragmented and uncoordinated at best. I came into post at the Queen Elizabeth Hospital Birmingham as a nurse specialist for renal metabolic disease in 2011. The role of a renal metabolic nurse specialist is to coordinate and manage the care and services provided to this patient group. The renal metabolic service aims to provide patients with the best care by conducting dedicated one stop clinics, providing psychological support, and empowering patients through the provision of patient information and education.

Some of the rare diseases managed affect multiple parts of the body. Consequently, people with these conditions require complex care from a range of different health professionals input which previously required the patient to make multiple trips back and forth from the hospital.

The nurse specialist role, acting as a care coordinator, has allowed us to develop one stop clinics which have been very well received by patients. The ability to coordinate care appropriately has impacted upon patients on a national level with many patients from all over the UK choosing to travel long distances to receive coordinated care so as to ensure they are receiving the best service..

Traditionally it was not uncommon for patients to feel angry and frustrated with the care they received due to the lack of understanding and knowledge around their condition. The presence of a nurse specialist has allowed patients to access a person who understands their condition to discuss their care and answer any questions or concerns they have and thus allowing patients to take an active role in their healthcare.

We have received extremely positive feedback from patients who have reported feeling increasingly reassured and confident in their health and no longer worry about whether or not they are receiving the most appropriate treatment.

It is my opinion that having a health professional available to coordinate care prevents patients from attending accident and emergency departments because

they have access to a health professional to act on their behalf with the ability to deliver the appropriate action or treatment at the most appropriate time. I believe that patients with rare renal conditions need access to care coordinators for them to be able to receive the best possible care. Without someone to coordinate their healthcare, it is inevitable that patients will suffer, and feelings of anger and frustration will prevail due to services being unable to meet the level of care required for a complex patient group.



Conclusions

There is a strong case for investment in care coordinator posts.

Evidence strongly indicates that having professionals in place who can fulfil the care coordinator role would be an effective and cost-effective way of providing care for people with rare diseases.

This report has demonstrated the potential cost savings that care coordinators can deliver, in the order of tens – or even hundreds – of thousands of pounds. Improvements in care and patient experience of care have also been shown where care coordinator posts are in place, and feedback from patients and families is extremely positive.

Care coordinator support must be provided in the most appropriate way to meet patients' and families' needs.

Research by RDUK indicates that patients and families do not mind who fulfils the care coordinator role, provided that someone is available to do so and that professional has sufficient knowledge of the condition and the situation.⁶¹

However, support for people with one rare disease may differ markedly from support needed by those with another rare disease. At the same time, certain groups of rare conditions may require similar support. Flexibility is therefore important in moving forward with the provision of care coordinator support, in order to meet the needs of all patients and families.

Funding for care coordinator posts must be adequate and sustainable.

It is essential that the level of investment in care coordinator posts is sufficient to provide support for those who need it. It is a concern that where care coordinator roles are already in place, there are not enough of them to ensure equitable access for patients, as evidenced by the examples in this report. RDUK also echoes the Royal College of Nursing's concerns that many specialist nurses – who help to coordinate care – are facing funding challenges.⁶²

The potential for joint funding of care coordinator posts could be explored. For example, DEBRA, the patient organisation for people with the rare skin condition Epidermolysis Bullosa (EB) part-funds EB nurses to provide specialised care, while core nursing activity is NHS-funded.⁶³

However, while in many cases patient organisations in partnership with health services can effectively provide care coordinators, such partnerships are not a solution for all rare diseases. Many patient organisations have the skills and expertise to fulfil this role, but lack the financial resources. Furthermore, most rare diseases do not have specific patient organisations, or the patient organisation may be too small and lack the capacity to undertake this role.

Ultimately, care coordinator posts need sustainable funding, which could pay for itself by generating savings, as demonstrated in this report. Without such funding, it is vulnerable patients who may be placed at risk.

61. 'Experiences of Rare Diseases: An Insight from Patients and Families', Rare Disease UK, December 2010

62. 'Specialist nurses – Changing lives, saving money', Royal College of Nursing, February 2010

63. 'Making the difference', DEBRA, 2012



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