REDUCE, IMPROVE, EMPOWER

Addressing the shared needs of rare autoimmune rheumatic diseases

RAIRDA
Rare Autoimmune Rheumatic Disease Alliance

FEBRUARY 2018

1. RAIRDA

Foreword

"This report is about the shared experiences of people living with rare Autoimmune rheumatic diseases, the impact of these conditions, and their unmet needs in accessing timely and effective healthcare.

Although focusing on three conditions, systemic lupus erythematosus, scleroderma and vasculitis, the results are likely to be applicable to other similar conditions, where the body's own immune system becomes overactive. As a result, this "friendly fire" then attacks healthy tissues, not just in one organ, but often throughout the body.

There is currently no cure, and all these conditions can significantly reduce the quality and length of life; for example, 1 in 7 people with systemic (ANCA-associated) vasculitis die within a year of diagnosis.

As a group, in contrast to the vast majority of all other rare diseases, these conditions predominantly occur in adult life, and are not primarily caused by genetic problems. We have therefore been concerned about whether the voice of these rare non-genetic autoimmune diseases has sufficient profile in the UK Strategy for Rare Diseases, and its implementation.

To address this, we have formed a new alliance, RAIRDA, to raise awareness of these similar needs, and support concerted action to resolve them.

This powerful report demonstrates for the first time the similar needs and experiences of people living with rare autoimmune diseases. It speaks very clearly of the shared needs to reduce diagnostic delay in all stages of the pathway, to improve care coordination across specialties, and of the pivotal importance of empowering clinicians and patients with greater knowledge.

Whilst our unique alliance between professional society and patient charities in this disease area has an important role in identifying and supporting solutions, their implementation will require concerted action across the community. We therefore need your support, whether you are a clinician, commissioner, policy maker or patient.

Thank you for reading this report, which we hope will catalyse further progress to improve the lives of people living with rare autoimmune rheumatic diseases."

Dr Peter Lanyon, Chair, Rare Auto-immune Rheumatic Disease Alliance

CONTENTS

1. RAIRDA 3

1.1 Introduction 6
1.2 Current landscape 7
1.3 About RAIRDA 8
1.3.1 Our vision 8
1.3.2 Aim and goals 8

2. ABOUT THE CONDITIONS 9

2.1 Lupus 10
2.2 Scleroderma 11
2.3 Vasculitis 12

3. SURVEY RESULTS 13

3.1 Diagnostic pathways 14
3.1.1 Diagnostic delay 14
3.1.2 Access to secondary care services 15
3.1.3 How to reduce diagnostic delay 16

3.2 Access to treatment 17
3.2.1 Fragmented care across multiple providers 17
3.2.2 Travelling for treatment 17
3.2.3 How to solve the problem 18

3.3 Coordination of care 19
3.3.1 Role of primary care 20
3.3.2 Specialist nurses 21
3.3.3 How to solve the problem 21

3.4 Wider impacts and additional support 22
3.4.1 Mental health 22
3.4.2 Physical impacts 23
3.4.3 Unplanned hospital visits 23
3.4.4 Work life 23
3.4.5 Sources of information 24
3.4.6 Disease guidelines 24
3.4.7 Additional support 25

4. RECOMMENDATIONS 26

References 27

KEY FINDINGS

44%

indicated that they waited more than three years from first symptom to receiving their correct diagnosis.

33%

of respondents routinely visit two locations for their care, with another third visiting 3 or more sites as a matter of course.

93%

of respondents reported seeing clinicians from multiple medical specialisms as part of their routine treatment. Just one in five are able to see multiple specialists at a joint clinic.

61%

of respondents were not confident that there was a coordinated plan in place for their care.

61%

of respondents state that they are struggling to cope with their condition.

44%

of respondents feel that their condition has had a negative effect on their family.

45%

of respondents reported that either they or their partners had reduced their working hours – or been forced to give up work entirely – as a result of their condition.

40%

say that they don't feel that they have enough information and support from the hospital in living with their condition.

1.1 Introduction

About 1 in 17 people will be affected by a rare disease at some point in their life – in the UK, that amounts to approximately 3.5 million people. The UK Rare Disease Forum highlights that 80% of rare diseases are genetic in origin, usually with onset in early life. The remaining 20% are non-genetic, an important subcomponent of which are the rare autoimmune rheumatic diseases. This term includes several multisystem conditions that share overlapping clinical and serological features, affect multiple organ systems, and therefore require coordinated multidisciplinary care. Examples include systemic sclerosis (systemic scleroderma), systemic lupus erythematosus (lupus), systemic vasculitis, Sjögren's syndrome, and myositis. These conditions can cause severe inflammation in different parts of the body, often simultaneously, leading to tissue or organ damage, which can be fatal. They predominantly have onset in later life (although can present during childhood), disproportionately affect women, and are not primarily of genetic cause. These distinctions between the rare autoimmune rheumatic diseases and the majority of rare conditions are important to make, not least to ensure parity.

In 2013, the UK Strategy for Rare Diseases was published, which for the first time since the establishment of the NHS, provided direction for the healthcare needs of people living with rare diseases. The strategy was well received and signed by the health ministers of each country in the UK, with cross-party support; it contains 51 distinct commitments that are designed to improve health and social care for rare diseases.

There is however, a concern that the focus of the strategy and implementation to date has been on rare genetic conditions, with a worrying lack of focus on the 700,000 or so people living with non-genetic rare diseases.

Four years on from the publication of the strategy, this lack of focus on rare autoimmune diseases means that there is a risk that the needs of people living with these conditions are being left behind.

To explore this further, in 2017, the Rare Autoimmune Rheumatic Disease Alliance (RAIRDA) conducted a survey of their patient members, designed to assess their experience of specific aspects of care highlighted by the UK Strategy for Rare Diseases. The survey was sent to people across the UK who are living with a rare autoimmune rheumatic disease. It was distributed via social media, e-newsletters and forums, had 47 questions and received more than 2,300 responses from across all four nations.

1.2 Current landscape

Following a European Union Council recommendation on rare diseases in 2012 the four countries of the UK jointly embarked on a consultation to develop a UK Strategy for Rare Diseases. The consultation outcomes informed the development of the UK Strategy, published in November 2013. To deliver the commitments in the Strategy, each of the four home countries agreed to develop their own implementation plan to reflect local health system arrangements and plans for the implementation of improvements. In Scotland, Wales and Northern Ireland, those implementation plans were introduced in autumn 2015, however the implementation plan for England was not published until this year, almost 5 years after the launch of the strategy.

While rare diseases have received greater attention since the UK Strategy was first published, diagnosis and delay remains a major challenge for the healthcare system, as does the coordination of care. Following the implementation of specialised commissioning for rare diseases by NHS England in 2013, although specialised centres have been identified, the number of these centres varies between regions, which can make geographical access and travel time a problem.

Currently, there is no coordinated national process to ensure comprehensive governance of the management of rare autoimmune rheumatic diseases or to support a cohesive drive to improve outcomes. As a result, there is likely to be significant variation in standards of care and outcome depending on where patients are treated. This tends to be influenced by both the process of care (e.g. within designated specialised as opposed to general clinics) and the degree of availability, support and interaction with specialised centres, where larger volume care, usually combined with research, is delivered.[1]

The need to improve the care for people with rare autoimmune diseases, catalysed by the UK Strategy, including the low visibility within this of the needs of non-genetic rare diseases, led the British Society for Rheumatology to host a national rare autoimmune disease workshop in November 2015. This brought together a range of stakeholders to help raise the priority of rare rheumatic and musculoskeletal conditions and improve patient care. The workshop and subsequent workshop report, ‘a collaborative approach to improving outcomes in rare rheumatic and musculoskeletal diseases’, and the UK strategy, were catalysts for the formation of a new alliance - RAIRDA.

1.3 About RAIRDA

In June 2016, LUPUS UK; Scleroderma & Raynaud's UK (SRUK); Vasculitis UK and the British Society for Rheumatology (BSR) came together to establish the Rare Autoimmune Rheumatic Diseases Alliance (RAIRDA), as an umbrella body for organisations with an interest in rare autoimmune rheumatic diseases. In 2017, the British Sjögren's Syndrome Association (BSSA) became an affiliate member, joining as a full member in 2018.

RAIRDA enables a united dialogue on areas of commonality (e.g. symptoms, treatments, diagnostic pathways, unmet needs and challenges) whilst at the same time enabling specific condition-related outputs. This alliance formally brings together for the first time, a dedicated body linking both clinical and patient organisations, and other key stakeholders, to improve care for people living with these conditions. We exist to provide a single, strong voice that will raise the profile of this group of conditions, influence policy and guide future research.

1.3.1 Our vision

People living with a rare Autoimmune rheumatic conditions, such as lupus, scleroderma and vasculitis, should receive the same focus and priority as other rare diseases.

1.3.2 Aim and goals

The aims of RAIRDA is as follows:

1. To raise the profile of the needs of people living with rare autoimmune diseases and their access to timely effective treatment

2. To promote the implementation of best practice care and pathways

3. To increase knowledge about patient care through better data

Further information on how RAIRDA intends to deliver these aims and address the issues raised in this report can be found here:[2] https://www.rheumatology.org.uk/Portals/0/Policy/RAIRDA/RAIRDA%20Strategy.pdf?ver=2017-10-26-150050-643

CASE STUDY

Alex was diagnosed with dermatomyositis and polymyositis in 1989, which then eventually evolved into a diagnosis of scleroderma in 2005.

"I've had extensive surgery on my wrists, hands, neck and shoulders with a lot of metal fusions to ease pain and deterioration. The scleroderma has affected my lungs quite severely, and I am now on oxygen."

2. ABOUT THE CONDITIONS

The three conditions covered by this report, lupus, scleroderma and vasculitis are lifelong conditions which currently have no cure. They develop when the body's own immune system starts to attack healthy parts of the body, leading to inflammation and damage in tissues or organs, which is often irreversible, and can be fatal. These conditions can affect many parts of the body (including joints, skin, lungs, kidneys and heart) and therefore often require similar cross-specialty medical expertise. Treatments usually involve "suppressing" the immune system with steroids and immunosuppressant drugs, each of which can cause serious side effects. However, in common with many other rare conditions, treatment options are limited; for example there has been only one new licensed drug for lupus in the last 50 years. Severe manifestations often need high-cost drugs, and there are commissioning policies for their use in each condition.

In addition, for many people living with these diseases, the effects may not be outwardly apparent, including nerve damage, kidney disease, and chronic pain. Many suffer from severe fatigue (as a result of both disease and treatment), and their health can vary from day to day making it difficult to meet the demands of a job and employer. Similarly, personal and social life can be devastated, resulting in breakdown of relationships.

2.1 Lupus

Lupus is a multi-symptom disease that can affect almost any part of the body. There are two main types: systemic lupus erythematosus and discoid lupus that affects the skin. Published estimates of the number of people in the UK with lupus are between 25 - 96 per 100,000, varying according to method of case ascertainment, gender and ethnicity, with more recent higher estimates possibly reflecting improved survival. The majority (80%) of people living with lupus are women, and the condition is more common amongst people of Black, Asian or Chinese ethnic origin. Lupus is not contagious.

25-96

cases of lupus per 100,000 people.

In lupus, the immune system produces autoantibodies that attack the body's own tissue. What causes this is not entirely clear, but appears to be due to a combination of environmental, hormonal and genetic factors. Two major symptoms of lupus are fatigue/extreme tiredness and joint and muscle pain. However, there are many others including: rashes, depression, anaemia, feverishness, headaches, and possible hair loss, mouth ulcers and kidney disease. Unless there is skin involvement lupus is effectively invisible

As symptoms can vary so much and also present at different times, diagnosis of lupus can be difficult and invariably involves a specialist and includes an array of blood and other tests, including ANA, Anti-dsDNA, CRP, ESR and urinalysis, along with a physical examination. The earliest possible diagnosis is important so that treatment can commence on order to reduce damage and to improve quality of life.

Lupus is treated with a range of medications, including non-steroidal anti-inflammatory drugs such as aspirin and ibuprofen, anti-malarials, commonly hydroxychloroquine, steroids. Immunosuppressants and biologics. Despite improvement in survival over the last 40 years, lupus patients still die on average 25 years earlier than the mean for women and men in the UK. Lupus is lifelong as there is presently no cure so the aim of treatment is to keep the disease under control using the minimum effective dosage of medication to keep side effects to a minimum.

2.2 Scleroderma

Scleroderma is a chronic and complex multi-organ disease. There are two main types of scleroderma: localised (affects mainly the skin) and systemic (internal organs are affected as well as the skin). With systemic sclerosis, the heart, oesophagus, blood vessels, kidneys, lungs, and digestive system can all be involved. Scleroderma affects approximately 20 in every 100,000 people.

There are three aspects to the disease processes: small blood vessel damage, autoimmune inflammation and tissue scarring. Scleroderma develops due to changes that occur in the body's connective tissues that lie under the skin and around the internal organs, which leads to too much fibrous connective tissue that is similar to scar tissue that forms after an injury. This scar tissue contains too much collagen that can cause the body's tissues to stiffen and thicken. The cause of scleroderma is unknown although an overactive immune system which attacks the body tissues is thought to be due to a mix of genetic and environmental factors.

Scleroderma symptoms vary widely from person to person and can include hardening/thickening of the skin, swelling of the hands and feet, heartburn and problems with swallowing, joint pain and stiffness, and blood vessel damage. Systemic sclerosis can also be difficult to diagnose as it can develop gradually and may take different forms. There is no single test. Like lupus, the diagnosis is made after taking into account a person's complete set of symptoms, along with a physical examination and blood tests.

Treatment will depend on the type of scleroderma and might include pain relievers, blood pressure medications that dilate blood vessels, medications to relieve the symptoms of acid reflux, medications to prevent infections and immune-suppressants. Life expectancy for people with scleroderma has improved, however it can be fatal if there is severe lung, heart and kidney involvement.

20

cases of scleroderma per 100,000 people.

2.3 Vasculitis

Vasculitis is a collective term for a group of autoimmune diseases involving inflammation of the lining of the blood vessels. There are 18 varied types of vasculitis, defined by the size of vessel affected. Reliable statistics on prevalence for rarer types of vasculitis are not available. Research suggests that the prevalence of ANCA vasculitis is 26 per 100,000, with an annual incidence of 1200 new cases nationally. Most types of vasculitis are potentially fatal if not promptly recognised and treated. The current understanding for vasculitis is that some individuals have a genetic predisposition to developing an autoimmune disease, but it also requires exposure to an environmental trigger (such as an infection, chemical or drug) to make the disease develop. The nature of the trigger may influence the type of autoimmune disease or the type of vasculitis.

26

cases of vasculitis per 100,000 people.

Symptoms of vasculitis vary widely, according to the type of vasculitis and the organs involved. Symptoms are often quite vague and non-specific, occurring over a long period of time, such as general malaise, fatigue, loss of appetite, joint and muscle pain, gastric pain, sinusitis, severe cough, loss of hearing, severe headaches, or loss of vision.

There are no specific tests for vasculitis although many tests may be done to help diagnose the condition. Blood tests to measure inflammation are useful but non-specific indicators of general inflammation. Some types of vasculitis rely heavily on imaging (X-ray, CAT, MRI/MRA scans), while biopsies can be an extremely useful diagnostic tool.

Treatment of vasculitis depends very much on the type of vasculitis and on the stage of the disease. In all cases, glucocorticoids (steroids) play a vital role in initial control of the disease. Traditional treatment usually involves an induction phase using a powerful chemotherapy drug, cyclophosphamide, to bring the disease under control. In some cases, such as where kidneys or lungs are severely involved, plasmapheresis (a type of dialysis) is used as an emergency measure to remove harmful antibodies from the blood. Once the induction drugs have brought the disease under control, more moderate immune suppressing drugs are used to maintain control. These drugs may be required over many years. In more recent times, a new generation of biologic therapies – targeted monoclonal antibodies - are now used in some cases, providing great benefit. However, the rarer types of vasculitis lack evidence for their use.

The most severe types of vasculitis can be life threatening and early diagnosis and treatment is essential to avoid permanent damage to tissues and organs. The outcome depends on the type of the disease and how it affects the individual.

3. SURVEY RESULTS

This report provides a description and analysis of our findings, along with recommendations that we believe will lead to improvements in the care and treatment for people living with these specific conditions.

2389 responses were received, of which 288 were excluded because either the diagnosis was not stated or could not be classified into one of the 3 main conditions. 2101 responses were analysed comprising people with lupus (1098), vasculitis (527) and scleroderma (476). The answers to each survey question were cross-tabulated according to diagnosis, and reported as frequencies and percentages. The results include all participants who answered the specific question being analysed, with no attempt to impute missing answers to questions. Where the answer to the survey question included an option for "Don't know", the analysis of people we report as having or not having the outcome of interest (e.g. time between symptoms and diagnosis) only included the people who were able to report an answer to the question.

Participants' demographics were representative of these cohorts, including differences between conditions. Two thirds were aged >45 years. Overall 90% were female, with, as expected, more males amongst people with vasculitis (22%), and more non-white respondents amongst people with SLE (9%). 81% of the respondents lived in England, 11% in Scotland, 3% in Northern Ireland and 6% in Wales, which is broadly representative of the population distribution of the UK.

3.1 Diagnostic pathway

Rare diseases, by their nature, frequently involve a longer diagnostic pathway than more common conditions, and are usually diagnosed in secondary, rather than primary care. It has been estimated that, from noticing the first symptom, the average rare disease patient consults with five doctors, receives three misdiagnoses and waits for up to four years before they receive their ultimate diagnosis.[3] This difficult diagnostic pathway is widely acknowledged and often referred to as the 'diagnostic odyssey.'[4]

3.1.1 Diagnostic delay

As with all conditions, rapid diagnosis is key to effective medical care and treatment. A delayed diagnosis can mean having to live with prolonged pain, with the untreated disease progressively worsening. A delay in accessing effective treatment also increases the risk of permanent organ damage and disability, and even death.[5] Delays limit access to early intervention programmes, mean greater waits for appropriate treatment, may result in patients receiving the wrong treatment in the interim,[6] and, often, means that multiple opportunities for timely interventions are missed.

Amongst the 95% of respondents who were able to report on their delay, almost half (46%) indicated that they waited more than three years from first symptom to receiving their correct diagnosis, with fewer than 1 in 10 being diagnosed in less than three months. As expected, there was some variation between conditions; people with lupus faced the longest delays, with (58%) reporting that it took more than 3 years from first symptom to diagnosis. Likewise, 40% of people with scleroderma reported a wait that long.

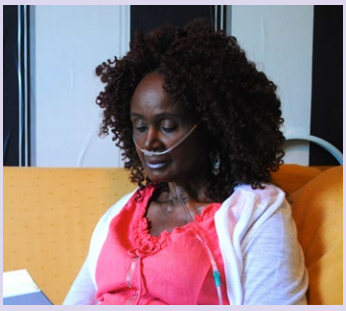
Amongst people with vasculitis delays were shorter, with more than half (58%) diagnosed within a year. However, this difference is likely to be related to the more acute presentation of vasculitis, which often presents with sudden organ damage (e.g. loss of vision, kidney failure), and where any delay in treatment carries risk of irreversible organ damage. Even so, 1 in 4 people with vasculitis (25%) report having to wait more than three years before receiving their correct diagnosis.

Interestingly, and of great concern, this delay does not appear to have improved in recent years. Amongst those diagnosed only in the last 3 years (approximately one third of all respondents), their diagnostic delays were very similar to the overall findings, with 42% waiting more than three years for their diagnosis. In comparison, 94% of cancer patients are seen by a specialist within two weeks of urgent GP referral for suspected cancer.[7]

46%

waited more than three years from first symptom to diagnosis.

CASE STUDY



Grace, aged 57, has had symptoms of scleroderma since the age of 33, when she was diagnosed with Raynaud's. It was another ten years before she was diagnosed with scleroderma.

"It was after quite a traumatic birth with my son that I started to notice symptoms, my skin was getting tighter, I had severe reflux and I began to struggle with everyday tasks. After continued visits to my GP, I was diagnosed with rheumatoid arthritis and put on various painkillers, which obviously didn't work.

My breathing was getting worse and I was having repeat visits to A&E. Thankfully on around my eighth trip to A&E, I met a young doctor who noticed scarring on my lung x-rays. I was given an immediate referral to a lung specialist where I was diagnosed with scleroderma. On diagnosis, I started the maximum course of the chemotherapy, to stabilise the aggressive nature of the disease.

Due to late diagnosis the extent of my disability is huge, as my lungs, skin, heart, kidneys and oesophagus are all affected. I am on oxygen due to my severe lung disease but I still feel constantly breathless."

3.1.2 Access to secondary care services

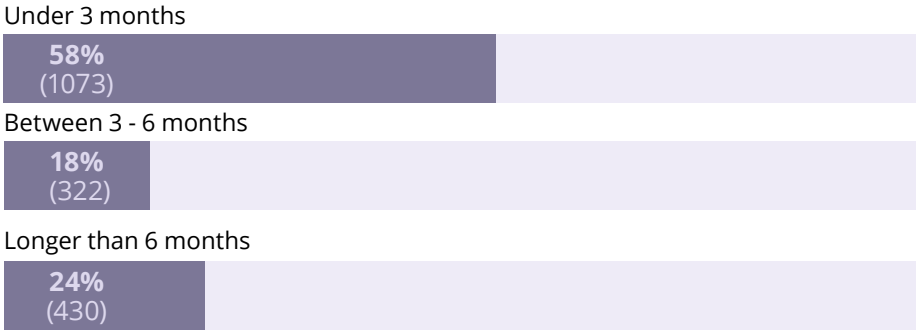
Patients with rare autoimmune rheumatic diseases often require assessment from multiple medical specialties, depending on the extent of organ involvement. This frequently includes rheumatology, nephrology, respiratory medicine, dermatology, ear, nose and throat, and ophthalmology. Coordinated access to the most appropriate specialist is a vital stage in the diagnostic pathway.

In England, the Department of Health's general target for non-emergency pathways is that 92% of patients should wait no more than 18 weeks from GP referral to seeing a specialist,[8] and that no patient should wait more than 52 weeks. In Scotland, the target is that 90% of planned or elective patients should commence treatment within 18 weeks of referral.[9] In Wales, that target is 26 weeks, with the aim that by March 2018, 75% of patients should wait no longer than 9 weeks for a diagnostic test, with no patient waiting longer than 26 weeks.[10] In Northern Ireland, the new 2017 Department of Health target is that by March 2018 no-one should wait more than 52 weeks for a first outpatient appointment.[11]

Among respondents, we can see there is a wide range of experiences of this wait. In total, just over half (54%) were seen by a specialist in under three months, while almost a quarter (22%) reported that they had waited longer than six months (26 weeks) for their specialist appointment. As with time to diagnosis, these results showed no significant improvement in these waiting times among those diagnosed most recently.

Our data suggest that current waiting time targets continue to be missed for people with rare autoimmune rheumatic diseases. Additionally, there is real concern that these targets themselves do not adequately reflect the need for prompt diagnosis of rare diseases to reduce the risk of irreversible organ damage occurring prior to treatment.

Time to see a specialist



CASE STUDY

Lisa died at age 38 from vasculitis, only 2 months after developing her first symptoms. She was admitted to hospital with severe joint and muscle pain, and rapidly became bed bound. Sadly, her condition deteriorated, and despite involvement of multiple specialists in her care, no definitive diagnosis was reached. She was discharged from hospital on steroid medication but was readmitted with pneumonia, when it was also discovered that vasculitis had destroyed the blood supply to her bowel, and she died of multi-organ failure.

3.1.3 How to reduce diagnostic delay

For rare rheumatic diseases, delays occur both in presentation to health professionals and in delayed recognition amongst healthcare professionals in both primary and secondary care.[12] There is an understandable lack of specialist knowledge of rare conditions among GPs that can result in delayed recognition, and longer time to referral and diagnosis, or to incorrect referrals to wrong specialties.

The UK Strategy for Rare Diseases included the recommendation that high quality training – including better training on rare diseases – be included on university courses (both undergraduate and postgraduate) as well as in professional development at work. It also calls for a common protocol to be developed to identify clear pathways for patients so that they receive a focused and coordinated diagnostic service.[13]

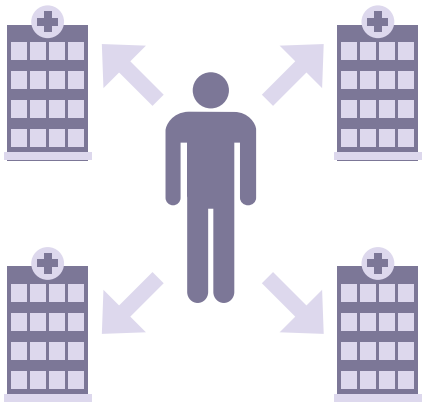
Recommendations: Reduce diagnostic delay

1. **The RCGP and RCP should seek opportunities to raise awareness of the importance of rare disease care, including equity of access regardless of geography, as a means to deliver recommendations from the UK Strategy for Rare Diseases and reduce diagnostic delays across the UK.**
2. **NICE should develop Quality Standards for rare autoimmune rheumatic diseases, including access and referral targets, leading on from the BSR's NICE-accredited guidelines for these conditions.**
3. **NHS England should formally assess the geographic availability and uptake of newer diagnostic tests (e.g. extended autoantibody screening tests, capillaroscopy, PET-CT) which might enable earlier diagnosis in undifferentiated/early cases of rare autoimmune rheumatic diseases.**
4. **All hospitals providing care for rare autoimmune rheumatic diseases should identify a relevant specialty clinical lead to coordinate local strategies for earlier diagnosis and ongoing care.**

3.2 Access to treatment

The challenge of managing a rare condition can be exacerbated by difficulties in accessing the correct treatment. Specialised centres for rare disease treatment are commissioned by NHS England, and are a source of additional expertise, although not all patients access care at these centres.

One third of respondents routinely visit 2 or more different hospitals to get the care they need.



3.2.1 Fragmented care across multiple providers

Although specialised centres providing multispecialty care can optimise the care pathway of patients with rare diseases, they have some inherent problems. The specialised centre model can also create a need for extensive patient travel, adding time burdens and an additional financial cost to people living with rare diseases that is rarely subsidised. Additionally, many of these conditions are poorly suited to such travel, due to mobility issues, lack of energy, or balance problems, all of which can turn long distance travel into an ordeal, and necessitate extra assistance or care.

Perhaps because of these problems, and despite the benefits, a Rare Disease UK study[14] has previously shown that only a quarter (27%) of patients with rare diseases are cared for in specialised centres. The vast majority of care for patients with rare diseases is therefore delivered close to home, relying on accessible services commissioned at a local level.

Lack of coordination can create a fragmentation of care delivery, with only a third (34%) of respondents to the survey receiving all their routine care in the past year at the same hospital. Another third (33%) routinely visit two locations for their care, with the other third visiting three or more sites as a matter of course. One in twenty respondents had visited five or more hospitals in the past year for their care.

3.2.2 Travelling for treatment

The secondary aspect of the centralised care/local care debate is travel time. Three quarters (75%) of respondents to the survey reported routine travel from their home to their hospital takes less than an hour, while less than 1 in 10 (8%) report a regular journey of two or more hours for their treatment. This would seem to reflect the expectations that many more people are receiving care in their local services than are travelling to a centralised, specialised centre.

Interestingly, when asked how far they would be prepared to travel to visit a specialist clinic, there appears to be a variation between respondents with different conditions – more than 1 in 4 people with vasculitis (28%) said they would be willing to travel over 100 miles to receive the specialist care they needed, while only 1 in 10 (13%) people with lupus would be willing to travel that far. That said, the wide spread of results suggests that, although many people with rare autoimmune rheumatic conditions would be willing to travel long distances in order to obtain specialised care, it is likely individual circumstance, rather than diagnosis, might be the key factor.

Respondents were also asked to indicate whether they had ever crossed a border to receive treatment. Amongst this small cohort, travel between Wales and England (in both directions) was the most frequent journey.

3.2.3 How to solve the problem

The UK Strategy for Rare Diseases states that specialist clinical centres should set a minimum standard of care for rare diseases and that each centre should:[15]

- have a sufficient caseload to build recognised expertise
- where possible, not depend on a single clinician
- coordinate care
- arrange for coordinated transition from children's to adult services
- involve people with rare conditions, and their families and carers
- support research activity

Where access to specialised centres is either not possible or not desired, it is essential that these standards of care are available to all people, regardless of geography, including care delivered across multiple sites. This can be supported by the development of coordinated networks between the specialised centres in each region and all local providers, with standardised protocols for care, and agreed thresholds for referral for specialised advice when needed, including the use of remote conferencing to facilitate this. Although there is an NHS England specialised rheumatology CQUIN to support the development of coordinated MDT clinics regional networks, there is very little information about the impact of these commissioning levers on improving care.[16]

CASE STUDY

It took Melanie three years to be diagnosed with systemic lupus erythematosus after the onset of her first symptom. Her condition also affected her brain (termed neuropsychiatric SLE), and she is under the care of a rheumatologist at one hospital, and a neurologist and a dermatologist at another, two hours apart from each other.

'I have been under the care of my local hospital for several years and have found all their rheumatologists to be excellent. I also see a neurologist and a dermatologist at another hospital. It was confusing for my GP and frustrating for my rheumatologist because the neurologists told me not to believe the rheumatologist and told me not to take the treatment. I didn't comply with treatment and became very ill again. My rheumatologist has looked after me very well and tried to coordinate my care between the two hospitals, but it was not nice for me, feeling like "piggy in the middle" between two hospitals and specialities. The neurologists have since sent me to their neuro-immunology specialist who has confirmed that I do have NPSLE.'

3.3 Coordination of care

The complex nature of rare autoimmune rheumatic diseases means that many patients will need to visit multiple specialists through the course of their treatment. The majority of respondents (1616/2101) were under the care of a rheumatologist, highlighting this specialty's role in the care of these conditions, and the great potential to lead on the coordination of care.

Number of people who report attending each specialty

The conditions covered by this survey are serious, life-long conditions, and each of them can have significant implications on lifestyle, mobility, and general wellbeing. With any condition involving multiple specialisms, coordination of care becomes a matter of significant concern. The UK Strategy for Rare Diseases notes that patients visiting different departments of the same hospital on different days – or, frequently, multiple hospitals – is rarely the best use of either the patient's time or the health service's resources.[17]

It is notable that almost a third (32%) of respondents feel that their care is not well co-ordinated. Again, there is some degree of variation between conditions, with lupus and vasculitis patients notably more likely to feel that their care lacks coordination than scleroderma patients. Even among those patients who felt that in general their care was well co-ordinated, more than half feel that there are still further improvements that could reasonably be made.

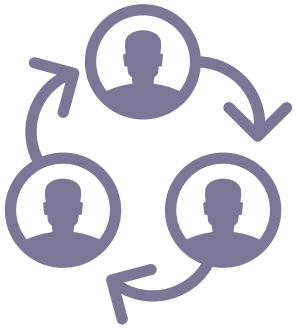
Identifying the underlying issues that hamper coordination of care is a challenge – the complex care involved with managing rare conditions mean that communication between different specialists is vital.

Figure 6: Percentage of people who report attending each specialty

Rheumatologist	80%
Dermatologist	17%
Cardiologist	16%
Ophthalmologist	15%
Nephrologist	14%
Respirologist	14%
Neurologist	11%
Orthopaedics	7%
Oral med	4%
Other	24%
No specialists	3%

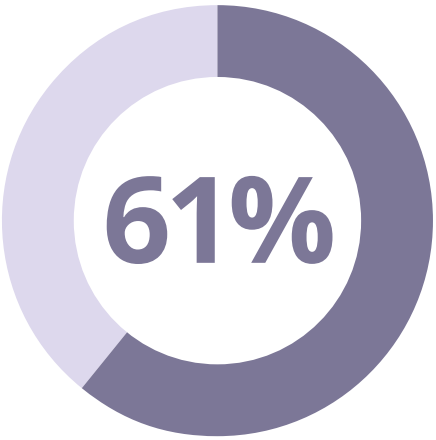
Overall, 93% of respondents reported seeing clinicians from multiple medical specialties as part of their routine care. Yet, among those people, less than one in five (17%) are able to see multiple specialists at a joint clinic. This figure is consistent across all conditions. Amongst those respondents who reported being under the care of both a rheumatology and respiratory specialist, only 20% were able to see them both at the same time in a combined clinic.

Perhaps most worryingly, respondents are far from confident that the various health professionals involved in their care have a plan for that treatment. Overall, just 39% of respondents feel that there is a coordinated plan for care in place, with some variation between conditions. Scleroderma patients (48%) and vasculitis patients (46%) are most confident in their plan for treatment. On the other hand, it is lupus patients who are most likely to feel that a care plan does not exist, with almost half (46%) stating that they don't feel the different professionals involved in their care have a plan for treatment. As of 2015, all patients with long-term conditions should have personalised care plans, according to the 2014-2015 NHS Mandate.[18]



93%

of respondents reported seeing clinicians from multiple medical specialisms as part of their routine treatment. Yet, among those people, less than **1 in 5** were able to see multiple specialists at a joint clinic.



of respondents are not confident that there is a co-ordinated plan for their care.

Although increasing the proportion of care delivered in combined specialty clinics is not a universal cure, the problems in care coordination are often exacerbated by administrative failures that result from a lack of a joined-up approach. Patients are sometimes transferred between multiple specialists in an attempt to find the right treatment, and management plans are more prone to suffer from errors that lead to frustration. From the health service's perspective, resources can be wasted both in terms of time, and the cost of prescriptions for treatments that prove ineffective. These issues, clearly, are not unique to rare diseases, but the nature of the conditions covered in this report inevitably make them a more common occurrence.[19]

3.3.1 Role of primary care

The specialised nature of drug treatment for rare autoimmune rheumatic diseases can result in primary care playing a secondary role in specific condition management for people living with these conditions. However, GPs are the prime carer for a person's overall health, including frequently being the first point of contact. Yet amongst our respondents, there is a widespread concern that GPs do not have sufficient knowledge about their specific conditions.

More than half (55%) of respondents reported feeling either not very or not at all confident in their GP's knowledge of their condition. This lack of confidence was highest among lupus and vasculitis patients (55% and 60% respectively), but even among scleroderma patients, who have the highest overall levels of confidence, just 12% describe themselves as being 'very confident' in their GP's knowledge. In contrast, 80% of cancer patients had confidence in their GP after they received their cancer diagnosis.[20]

of respondents reported feeling either not very or not at all confident in their GP's knowledge of their condition.

CASE STUDY

Amy, aged 24, has had symptoms of lupus since the age of 11, when she developed circulatory problems and was diagnosed with Raynaud's syndrome. It was another three years before a diagnosis of lupus was suspected, and it took a further two years for her diagnosis to be confirmed.

"I've perhaps always had signs of lupus, spending my childhood in and out of hospital with various things. When I was 11, my Grandma noticed a chilblain on my toe and got my Mum to take me to our GP. She diagnosed Raynaud's and referred me to our local rheumatologist. Over time, I was getting large severe rashes over my legs and body. I was referred to a specialist based in London, who after a year of seeing me 'diagnosed' lupus. I wasn't told in the appointment, I was approached by a nurse after the appointment who said 'are you the one with lupus?' to which Mum and I, confused, replied 'yes?'. I was admitted twice for treatment over a few years before I got a definitive diagnosis of lupus."

3.3.2 Specialist nurses

The use of specialist nurses to lead a coordination of a patient's care has become a more common practice in rheumatology services over recent years. Where this happens, a specialist nurse acts as a case manager to coordinate access to treatment and support across secondary, community and primary care for their patients. A study by the Royal College of Nursing on the work of rheumatology nurse specialists across the UK shows that they 'represent good value for money, through reducing costs in primary care and saving consultants' time.'[21] The Royal College of Nursing calculates that outpatient work by rheumatology nurse specialists is worth £72,128 per nurse, and saves the NHS as much as £175,168 per nurse each year by freeing up consultant appointments. Further savings are made through a reduction in GP appointments as specialist nurses make themselves available for telephone consultations with their patients.[22]

Among respondents to this survey, only one in three (37%) reported that their care is supported by a specialist nurse. This figure compares, for example, to 87% of people with cancer who are given the name of a clinical nurse specialist.[23] Given the identified benefits of access to a specialist nurse, this is a disappointingly low figure, and may suggest that further work needs to be done on ensuring access to this resource. For those who do have access to a specialist nurse, the majority (61%) say that their nurse is easy to contact, emphasising the beneficial role that the service can play.

3.3.3 How to solve the problem

The Department for Health acknowledges that coordinated care is essential when many specialists across many departments are seeing the same patients[24], but finding a solution to the problem has proven to be far from straightforward.

Increasing access to specialist nurses with specific high-level expertise in these conditions and increasing the prevalence of joint clinics are two actions that should have significant impact on the experiences of people living with rare autoimmune rheumatic diseases. Realising these two goals, however, will rely heavily on local priority setting and budgeting, making any national approach to the problem difficult unless barriers to initiating combined clinics, in particular suitable tariff arrangements, are overcome.

Recommendations: Improve coordination of care

5. **NHS England should provide additional support for development of coordinated care and networks for rare autoimmune rheumatic disease by disseminating information about uptake, specific utilisation, and clinical impact of the relevant CQUINs, so that all clinicians and providers learn from this.**
6. **NHS England should accelerate plans for revisions to tariff, to ensure there are appropriate levers for the stable financial development of multi-specialty clinics in hospitals where there is sufficient clinical demand for this.**
7. **For each condition, a simple clinical "checklist", based on the BSR guidelines, should be developed, to help ensure that the most essential aspects of care are routinely delivered at each interaction, including assessment and control of disease activity and strategies for prevention of co-morbidities.**
8. **All hospitals should ensure that every patient with a rare autoimmune disease has a named person responsible for coordinating their care. There should also be rapid access to specialist advice during emergency and unscheduled care, facilitated by this need informing local job planning discussions.**
9. **RAIRDA should work collaboratively with NHS England and the Royal College of Emergency Medicine to explore the introduction of 'alert cards' to enable specific needs to be considered when emergency and unscheduled care is required.**

3.4 Wider impact and additional support

Alongside the physical impacts of the conditions covered in this survey, people living with rare conditions can face other significant difficulties.

3.4.1 Mental health

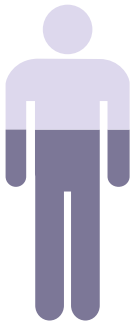
Uncertainty over symptoms, conflicting or incorrect information, false diagnoses and frequent trips to different doctors and specialists all serve to cause stress and anxiety in patients and can have significant impacts on work and home life.

The first Europe-wide survey on the social impact of rare diseases was published in May 2017,[25] and has given a new insight into the serious impact that rare diseases can have on everyday life, with over 80% of patients and families affected. Challenges of this nature are not regularly accounted for within the social care system, worsening the problems. Data suggests that, in the UK, it can take up to six years for a correct diagnosis of a rare disease. This is backed by our own survey, with almost half (46%) indicating that they waited more than three years from first symptom to receiving their correct diagnosis.

Diagnosis itself is all too often followed by more stress – be it difficult decision-making on treatments, frustration over a lack of options and means of managing the illness, or simply learning to cope with the impact of the condition. Finally, many of the conditions considered here have significant direct or indirect expenses, adding to the stress. These impacts are felt by patients and caregivers alike.

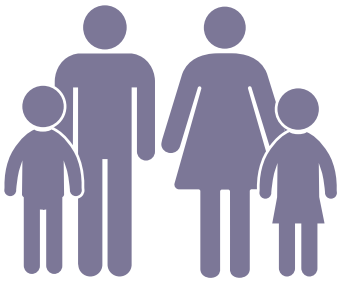
In total, 61% of respondents state that they are struggling to cope with their condition, with lupus patients facing the most difficulties. More than half (52%) of people say they feel isolated or lonely at least once a week as a result of the effects of their condition, with one in five (21%) feeling that way every day. As with the previous question, lupus patients are most likely to feel isolated, with 24% feeling that way every day, and 57% at least once a week.

Finally, 44% of patients also feel that their condition has had a negative effect on their family – with lupus patients (50%), again the most likely to have seen negative impacts.



61%

of respondents state that they are struggling to cope with their condition.



44%

of respondents feel that their condition has had a negative effect on their family.

CASE STUDY

Lorraine was diagnosed with scleroderma in 2010. The condition has caused insulting and hardening to her skin and underlying muscles leading to facial changes and clawing of the hands, affecting her confidence and mental health .

“My confidence has suffered, I feel ugly as my face has changed and I don’t like having to have my family help do the things I used to do. It has robbed me of some of the things I love to do like baking, as I find it hard to hold the utensils.

I sometimes think can go back to work on a good day but, then I wake up next day and I’m ill. So my mental thoughts are at a discourse with my body’s own capacity so that is very frustrating at times.”

3.4.2 Physical impacts

The majority of rare autoimmune rheumatic diseases have a significant physical impact on patients. Of those surveyed, just 4% reporting that feeling tired, weak or fatigued has not been a problem, with more than half of lupus (60%) and vasculitis (53%) patients report feeling those physical effects every day.

Worryingly, the data suggests that guidance on these impacts is lacking. Less than a quarter (23%) of respondents have received advice on the amount of daily physical activity or exercise they should do, and even fewer (13%) have received support on how to manage their body weight while living with their condition.

3.4.3 Unplanned hospital visits

Just under half (49%) of those surveyed reported having to make an unplanned hospital visit in the past year, with vasculitis patients the most likely to have been required to make multiple visits (34% of that group reported making 2 or more unplanned hospital visits). The majority (62%) reported that they are satisfied by the treatment they received on these visits.

3.4.4 Work life

While there is no clear pattern to the overall work status of respondents, with 24% employed full-time, 14% employed part-time, 13% unemployed, and 28% retired, there are some indicators of the impact of rare autoimmune rheumatic diseases on the ability to work.

Although just 15% of respondents to the survey are aged 66 or above, 28% of respondents are retired. While it should be noted that a further 23% of respondents fall into the age 56-65 range, and the majority (91%) of respondents are female (for some of whom statutory retirement age will have been earlier than 65), this might suggest that the conditions covered in this report are likely to force people to take early retirement in some cases.

Respondents who were employed, meanwhile, are about twice as likely to be working full-time (24%) as part-time (14%) – compared to the UK workforce as a whole, this suggests that people with one of the conditions covered in this report are more likely to work part-time, where the ratio of full-time to part-time work is approximately 3:1.[27] The unemployment rate (13%) among respondents is three times higher than among the UK population as a whole.

These findings are further supported as 25% of respondents reported that either they or their partner or carer had reduced their working hours as a result of their condition, and a further 20% reported that either they or a partner or carer had been forced to give up working due to their condition.

Among those who are still in work, one in ten report having missed more than three months of work in the past year because of their condition. Vasculitis patients are the most likely to have prolonged absences from work, with 16% having forced absences that long.

1 in 5 have missed more than 3 months of work in the past year because of their condition.

CASE STUDY

Jay is a former teacher who was Head of IT within a thriving school until her scleroderma became so severe that she was unable to continue with her career.

"Sadly, my full-time teaching career came to an abrupt end, when I started to experience a series of debilitating physiological symptoms ranging from extreme fatigue, cold hands, chronic pain and repeated respiratory infection, which were all symptoms of my scleroderma.

With chronic exhaustion and fatigue, I miss out on some amazing life events – it is devastating to see a whole day or week go past, when previously on these days I would have managed the IT education of up to 1000 children, team meetings and parents evenings."

3.4.5 Sources of information

The complex nature of rare autoimmune rheumatic diseases, and the varied impacts – including those beyond the direct symptoms – discussed in the previous section, mean that patients need complex support mechanisms, both within and outside the health service. Unfortunately, as with many rare conditions, wider understanding of these impacts is often lacking, leading to gaps in provision, and potentially worsening the feelings of isolation that many respondents report feeling.

Less than half (46%) of respondents report having access to a telephone advice line when they have a question about their condition, while over a third (40%) say that they don't feel that they have enough information and support from the hospital in living with their condition.

Top 3 most used sources of information

Internet search	**63%**
Consultants	**58%**
Online support forums	**48%**

Respondents were asked to indicate the various different sources of information that they found to be most helpful in gathering support for issues arising from their condition, or in finding out more about the condition itself. The most popular source of information was the internet (with 63% of people indicating they found it a useful resource) followed by consultants (58%), and online support forums (48%). These results likely reflect the ease of access to those resources – although a strong reliance on the internet as a source of information brings its own concerns, not least the risk of being exposed to incorrect or misleading advice and information.

Of the people who reported they had access to a specialist nurse, less than half (46%) indicated that they considered them to be a helpful source of information. Local support groups, and support group literature – that is physically available at clinics or GP surgeries – was not considered an important source of information by the majority of patients.

3.4.6 Disease guidelines

BSR produces NICE-accredited guidelines that cover diagnosis, assessment, monitoring and treatment of patients with specific conditions, and these are a vital tool in setting the standards of care for people with rare diseases. While primarily aimed at rheumatologists and clinical nurse specialists, they are also widely used by other specialist practitioners, emergency medicine practitioners, GPs and many trainees. There are BSR guidelines for ANCA vasculitis, scleroderma, and sjögren's syndrome. The lupus guideline, for example, addresses the care of patients with common symptoms such as skin rashes and arthritis as well as those with less common, but potentially more serious problems like kidney disease. It encourages the referral of patients with the most serious and difficult-to-control diseases to specialised centres with experience of new therapies and with multi-disciplinary team backup, thus promoting a coordinated networked care approach.

Implementation of guidelines can help reduce the current diagnosis times and result in earlier access to appropriate treatment, and so giving a better quality of life for patients. They can also play the secondary role of promoting awareness and knowledge of the condition for the patients themselves.

The data, however, suggest that the existence and purpose of these guidelines is not widely known outside of professional circles. The majority (78%) of respondents were unaware that a guideline exists for their particular condition, with vasculitis patients the most likely to be aware (35%). This is likely to be a reflection of the fact that BSR guidelines for vasculitis predated those for lupus, scleroderma and sjogrens by 9 years. Among those who were aware of the guidelines, there is a lack of certainty that treatment is matching the recommendations – 40% of respondents who are aware of the guidelines, say they do not know whether their treatment was in accord with that document.

3.4.7 Additional support

Given the obvious connections between physical and mental health, respondents were also asked whether they had ever been offered support from the NHS in dealing with the psychological impacts of their condition. Extremely worryingly, just 16% report that they are being offered such support. Also of concern, 1 in 10 respondents report that they had previously asked to receive psychological support, but that it had not been forthcoming, while a third of people who had received such support only received help after having asked for it.

Finally, we asked all respondents whether, in general, they feel well supported by the NHS in dealing with their condition. Although the majority (68%) said they are, a significant proportion – 26% - feel that they are not adequately supported.

1 in 10 respondents asked for psychological support but did not receive any help.

Recommendations: Empower with greater knowledge

10. Providers of care should ensure that people with rare autoimmune diseases are empowered by appropriate education and support from a specialised nursing and allied health professional multidisciplinary team, including emotional and psychological support.
11. Public Health England's National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) (and equivalent bodies in the devolved nations) should work with RAIRDA to facilitate identification and registration of people with rare autoimmune rheumatic diseases, including promotion of self-registration, to better identify their needs, outcomes and variation in care.
12. Further health-services research should be undertaken to assess what benefits have accrued for people living with rare autoimmune rheumatic diseases since 2013, as a consequence of the UK Strategy for Rare Diseases and introduction of Specialised Services Commissioning.

4. RECOMMENDATIONS

Reduce diagnostic delay

1. The RCGP and RCP should seek opportunities to raise awareness of the importance of rare disease care, including equity of access regardless of geography, as a means to deliver recommendations from the UK Strategy for Rare Diseases and reduce diagnostic delays across the UK.
2. NICE should develop Quality Standards for rare autoimmune rheumatic diseases, including access and referral targets, leading on from the BSR's NICE-accredited guidelines for these conditions.
3. NHS England should formally assess the geographic availability and uptake of newer diagnostic tests (e.g. extended autoantibody screening tests, capillaroscopy, PET-CT) which might enable earlier diagnosis in undifferentiated/early cases of rare autoimmune rheumatic diseases.
4. All hospitals providing care for rare autoimmune rheumatic diseases should identify a relevant specialty clinical lead to coordinate local strategies for earlier diagnosis and ongoing care.

Improve coordination of care

5. NHS England should provide additional support for development of coordinated care and networks for rare autoimmune rheumatic disease by disseminating information about uptake, specific utilisation, and clinical impact of the relevant CQUINs, so that all clinicians and providers learn from this.
6. NHS England should accelerate plans for revisions to tariff, to ensure there are appropriate levers for the stable financial development of multi-specialty clinics in hospitals where there is sufficient clinical demand for this.
7. For each condition, a simple clinical "checklist", based on the BSR guidelines, should be developed, to help ensure that the most essential aspects of care are routinely delivered at each interaction, including assessment and control of disease activity and strategies for prevention of co-morbidities.
8. All hospitals should ensure that every patient with a rare autoimmune disease has a named person responsible for coordinating their care. There should also be rapid access to specialist advice during emergency and unscheduled care, facilitated by this need informing local job planning discussions.
9. RAIRDA should work collaboratively with NHS England and the Royal College of Emergency Medicine to explore the introduction of 'alert cards' to enable specific needs to be considered when emergency and unscheduled care is required.

Empower with greater knowledge

10. Providers of care should ensure that people with rare autoimmune diseases are empowered by appropriate education and support from a specialised nursing and allied health professional multidisciplinary team, including emotional and psychological support.
11. Public Health England's NCARDRS (and equivalent bodies in the devolved nations) should work with RAIRDA to facilitate identification and registration of people with rare autoimmune rheumatic diseases, including promotion of self-registration, to better identify their needs, outcomes and variation in care.
12. Further health-services research should be undertaken to assess what benefits have accrued for people living with rare autoimmune rheumatic diseases since 2013, as a consequence of the UK Strategy for Rare Diseases and introduction of Specialised Services Commissioning.

References

1. IM 3 Auto-immune Management, NHS England 2016; Accessed online January 2018: https://www.england.nhs.uk/wp-content/uploads/2016/11/im3-auto-immune-management.pdf

2. https://www.rheumatology.org.uk/Portals/0/Policy/RAIRDA/RAIRDA%20Strategy.pdf?ver=2017-10-26-150050-643

3. The Rare Reality – an insight into the patient and family experience of rare disease, (Rare Disease UK 2016) accessed Dec 2017

4. Diagnostic odyssey for rare diseases: exploration of potential indicators, (Policy Innovation Research Unit 2015), accessed Dec 2017.

5. A collaborative approach to improving outcomes in rare rheumatic and musculoskeletal diseases.(British Society for Rheumatology 2016), accessed Nov 2017.

6. The UK Strategy for Rare Diseases, (Department of Health, 2013), accessed Oct 2017.

7. Department of Health, Social Services and Public Safety. Cancer Waiting Times. Accessed June 2015.

8. Consultant-led Referral to Treatment Waiting Times (NHS England 2017); Accessed Oct 2017 from: https://www.england. nhs.uk/statistics/statistical-work-areas/rtt-waiting-times/

9. 18 Weeks: The Referral to Treatment Standard (NHS Scotland 2008); Accessed Oct 2017: http://www.gov.scot/Resource/Doc/211202/0055802.pdf

10. NHS Activity & Performance Summary: November/December 2017 (NHS Wales 2017); Accessed Nov 2017

11. Department of Health, Northern Ireland Waiting Time Statistics: Outpatient Waiting Times Quarter Ending June 2017

12. A collaborative approach to improving outcomes in rare rheumatic and musculoskeletal diseases.(British Society for Rheumatology 2016), accessed Nov 2017.

13. The UK Strategy for Rare Diseases, (Department of Health, 2013), accessed Oct 2017.

14. Centres of Excellence for Rare Diseases (Rare Disease UK 2013) Accessed Nov 2017

15. The UK Strategy for Rare Diseases (Department of Health 2013); accessed online, Dec 2017;

16. National Programmes of Care and Clinical Reference GroupsInternal Medicine, A09 Specialised Rheumatology (NHS England)

17. The UK Strategy for Rare Diseases, (Department of Health, 2013), accessed Oct 2017.

18. The Mandate A mandate from the Government to NHS England: April 2014 to March 2015 (Department of Health 2013); accessed online, Dec 2017; www.gov.uk/government/publications/nhs-mandate-2014-to-2015

19. Rare disease perspectives 2016 (Findacure 2016); Accessed online, Nov 2017;

20. Patients' confidence in their GP before and after being diagnosed with cancer (Mette B Larsen et al 2011) accessed online, Nov 2017

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

22. Rare Disease Care Coordination: Delivering Value, Improving Services, Rare Disease UK 2013

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

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

25. Juggling care and daily life The balancing act of the rare disease community A Rare Barometer survey' Eurodis: Rare Diseases Europe 2017

26. Office for National Statistics: October 2017, available at: https://www.ons.gov.uk/employmentandlabourmarket/peopleinwork/employmentandemployeetypes/bulletins/uklabourmarket/october2017.

Acknowledgements

We gratefully acknowledge the assistance of Dr Fiona Pearce, Arthritis Research UK Clinical Research Fellow & Clinical Lecturer in Rheumatology, University of Nottingham, who analysed the survey data, and is the guarantor of the analysis.

We would also like to acknowledge the survey respondents and those who submitted case studies for use in this report.

While there are inherent limitations to survey data, including accurate recall of past events, survival bias and self-selection of participants, the age and sex distribution of respondents was as expected from the demographics of these diseases, as were differences between responses according to each condition. The strength of the survey results are the large number of respondents, and the opportunity for people with a broad range of experiences to contribute.

Authors and contributors

Jack Feinmann, Policy and Public Affairs Officer, RAIRDA

John Hopgood, Head of Policy and Public Affairs, BSR

Peter Lanyon, Chair, RAIRDA

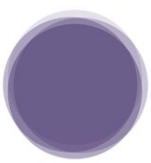
Sue Farrington, CEO, SRUK

Chris Maker, CEO, LUPUS UK

John Mills, Chair, Vasculitis UK

Miguel Souto, Director of Clinical Affairs, BSR

Ali Rivett, CEO, BSR



RAIRDA

Rare Autoimmune Rheumatic Disease Alliance