

Introduction to response from the Rare Autoimmune Rheumatic Disease Alliance (RAIRDA)

RAIRDA is a UK-wide coalition of four charities: Vasculitis UK, LUPUS UK, British Sjogren's Syndrome Association, and Scleroderma and Raynaud's UK, which calls for improvements in the way these rare diseases are cared for.

Rare autoimmune rheumatic diseases (RAIRDs) can be split in two groups: connective tissue disorders (lupus, scleroderma, myositis, primary Sjögren's syndrome) and systemic vasculitis (ANCA-associated vasculitis, giant cell arteritis, Takayasu's Arteritis and Behcet's disease). These conditions are characterised by the body's own immune system becoming overactive and attacking healthy tissues, often in multiple organs throughout the body simultaneously, leading to tissue or organ damage which can be fatal. These conditions can affect many parts of the body (including joints, skin, lungs, kidneys and heart) and often require cross-specialty medical expertise. Unlike the vast majority of rare diseases, these conditions predominantly occur in adult life and do not have a simple genetic link. They also disproportionately affect women, for example women make up approximately 80-90% of lupus patients. The impacts of these conditions on a patient can vary depending on the organ(s) affected but they can be severely life limiting and life threatening.

We used evidence from two surveys, UK-wide survey of over 2000 patients (125 of whom were Welsh patients) published in 2018 and a survey conducted in June 2021 with 126 responses from patients in Wales, to write this response.

Based on research using healthcare data in England which indicates there are 174,000 people who have ever been hospitalised with a RAIRD in that country, we believe there are around 9,000 people with these diseases in Wales.

We believe the evidence presented in this submission supports:

- The commissioning of a specialised centre, or specialised centres, for these conditions within Wales.
- Use of specialised networks, involving all departments which care for people with RAIRDs, to support local rheumatologists to care for these conditions, and make sure people get specialised care when needed.
- Urgent, and sustained, action to address workforce issues in rheumatology.
- Investment in the wider multidisciplinary teams including specialist nurses and clinical psychologists.
- Development of digital solutions to support monitoring tests being carried out closer to patients, with specialists still able to oversee these as needed.
- Ensuring access to rapid advice for issues like flares, such as through advice lines able to support people with rare diseases.
- Action to ensure face-to-face appointments are available when needed.
- Specific recommendations to support rare autoimmune rheumatic diseases, rather than generic recommendations covering rare diseases in general.

Overall

1. Question 1: In light of the effect of the pandemic on service, what should musculoskeletal services consider?

Telephone appointments are often not sufficient for people with rare autoimmune rheumatic diseases by themselves.

These appointments may be seen positively by patients when they do not need physical examination, or can have their monitoring tests done locally by other services, and discussed with their specialist during their phone consultation.

However, when telephone appointments mean that either symptom are not discussed fully or that monitoring tests are not done, they are insufficient for rare autoimmune rheumatic diseases.

A difficulty noted by clinicians across the UK is knowing which patients need to be seen physically and which can be seen remotely. Sometimes this means that a clinician only realises someone needs to be seen physically after a remote appointment. Often patients themselves are the best judge of this and, in an ideal world, patients would be able to choose a physical or remote appointment.

It is worth noting that even if no follow-up physical appointment is needed, remote appointments are not necessarily quicker or time-saving for clinicians.

For example, one patient in Wales told us:

“It has been hard to access sufficient emotional and psychological support and having on 2 yearly telephone discussions has exacerbated this as they focus on symptoms and medication”

Question 2: What digital solutions are required to support clinicians undertaking their role?

Remote monitoring tests can clearly be supported and require digital solutions. For example: tools such as digital platforms used to log blood tests done locally so that specialists can review them.

The development of networks for the delivery of specialised rheumatology care (discussed below) will work best with digital tools which allow collaboration, data collection and coordination across health boards.

Question 3: What extra provision is needed to support those on waiting lists or receiving management and support in primary care?

In June 2021 we asked patients what impact the pandemic had on the care they receive for their condition and the impact this had on their ability to manage their condition. 41% said it had a major negative impact, 29% said it had a minor negative impact.

There is now a substantial number of relatively stable patients who have missed routine tests for far longer than the recommended interval. This is due to the fact services have, understandably,

prioritised those patients who do not have stable disease or who were newly diagnosed.

In this prioritised group some patients recognised and welcomed how responsive consultants and hospitals had been despite the pressures of the pandemic, although their care had not been unaffected. However, this was not a universal experience, with some patients experiencing difficulties with getting in contact with their services.

“Whilst face to face appointments were swapped to telephone appointments during the pandemic when I had issues and new symptoms I was seen in clinic within the same week.”

“I went privately as I didn’t know who to contact during a flare. He explained it will be due to COVID that the rheumatology nurses don’t answer the phone, or reply to messages left”

“Getting access to phone calls with specialist nurse or rheumatologist has been incredibly difficult through the pandemic.”

“I had my rheumatology appointment cancelled during March 2020. I have not heard anything more from the hospital since. I am unwell & was hoping to start a new treatment for the conditions I've had diagnosed. I feel there is no help whatsoever and as if I have fallen off the radar completely. Just left in pain and forgotten about.”

“There is a need to be seen more quickly. Initial wait time for first appointment was very long and because of COVID appointments keep getting cancelled.”

We do think that the coordination and multi-specialty care needed for these diseases has compounded the disruption caused by the pandemic.

“The pandemic meant a delay to my heart scans and MRI scans. I still haven't seen my cardiologist and spoke with a registrar who claimed she would speak with my consultant, but the letter was signed off by someone who isn't my consultant. I don't trust them, and it's meant my care isn't collaborative. My renal appointments went from every 2-3 months to over a 12 month delay even though I have kidney problems as a result and high blood pressure. I haven't even heard from neurology and no idea if I'm still on their books.”

It is important to underline how important responsive care is. The ability of patients to get assistance early can help prevent issues escalating and patients requiring more care from the health service.

In addition to the need to be responsive to obvious needs, regular monitoring is important to help. 16 months into the pandemic, we are now concerned that the group of patients who are relatively stable, but still need monitoring tests to check for potentially asymptomatic issues, are struggling to access these tests.

“I have been satisfied with my care, generally, but feel that the pandemic has had a major negative effect on those patients who are considered as not requiring urgent treatment.”

Deprioritising these patients for a short time is justifiable, but the longer they are left without

monitoring, the greater the risk that they will start to require intensive and urgent care.

We would recommend making sure urgent shared care protocols are put in place to support primary care to do the basic monitoring tests, with specialists able to review results remotely.

In other areas of the UK where community blood testing has been implemented in response to the pandemic, this has been welcomed by both specialists and patients.

The longer waiting times caused by the pandemic will increase the time taken to diagnose patients, as well as making caring for existing patients difficult. New ways of working, as well as increasing the capacity of rheumatology departments, will be important to deal with this.

Delays to diagnosis will have a substantial impact on patients. We know that long diagnostic journeys cause serious distress and heighten the impact these conditions have on patients' mental health, but there is also evidence they increase the costs on health services in the long-run as disease is left uncontrolled, increasing the chance that it is able to cause serious damage to organs

“My consultant thinks I would have been diagnosed sooner had it not been for the impact of the pandemic on services. As a result, I was very ill when I was diagnosed, which has implications going forward.”

Introduction

Question 4: Does the introduction identify the key issues relating to the conditions and services throughout Wales?

We welcome the mention of rare autoimmune rheumatic diseases (RAIRDs) here and this is important to highlight the diversity of the conditions dealt with by MSK services.

We would suggest elaborating that these conditions affect internal organs, not just elements of the MSK system. For example, amend: The term ‘MSK conditions’ includes a wide range of health conditions affecting the bones, joints, muscles and spine, as well as rarer autoimmune conditions (such as systemic lupus erythematosus, Sjogren’s syndrome, scleroderma, and vasculitis) which can affect internal organs.

Actions

Question 5: Are the proposed actions appropriate and will they provide the service required?

Our key asks, for rare autoimmune rheumatic diseases are:

- A specialised, tertiary centre (or centres) for these conditions should be formally commissioned in Wales. Whilst some individual departments and clinicians do run dedicated clinics for these conditions, this is not the same as a service formally commissioned by WHSSC.
- A national network should be established to coordinate the care of these conditions between a specialised hub(s) and local rheumatology departments or ‘spokes’. Establishing this network well will require investment and ‘buy-in’ from every health

board involved in caring for RAIRDs, but will allow care which can be delivered locally, improve the quality of that care, and mean patients can have access to the specialised care they need.

- A network model should replace the system of Individual Patient Funding Requests, to facilitate shared care between specialist centres and local rheumatology departments across health board borders.
- These specialised services and the network should continue to support access to more specialist support in England when necessary.

This draft framework alludes to these changes, but, as we note in this submission, the lack of specific actions is disappointing.

A recent study using data from NCARDRs in England estimates that there are 174,000 people in England who live with rare autoimmune rheumatic diseases. We expect this means there are around 9,000 people in Wales with these conditions.

We think this draft framework misses an opportunity to give health boards specific, realistic, actions on rare diseases which link to the intentions laid out in pages 36-38, namely, the establishment of specialised services and networks to care for rare autoimmune rheumatic diseases.

Instead of referring to rare diseases in general, as recommendation 27 does, there should be inclusion of actions specific to rare autoimmune rheumatic diseases, given these are the specific rare diseases which rheumatology services often deal with.

We are concerned that there is no reference to the new UK Rare Disease Framework, or the action plan the Welsh Government is drawing up in response to this in the list actions. The Rare Disease Strategy is now being replaced by the Rare Disease Framework. Given the time period this MSK framework will be in effect for, the Rare Disease Framework should be referenced. It is vital that thinking is joined up across different policies. Given the UK Rare Disease Framework calls for action on non-genetic rare diseases, we believe this MSK Framework should contain specific references to RAIRDs, as a significant group of these diseases.

Wales Rare Disease Strategy Implementation Plan (2017, page 15, action 4) noted Wales would: *Work with health boards as providers and NHS England to:*

- *develop and agree service specifications;*
- *develop an agreed process for monitoring services against specification; implement and monitor services against service specification including outcome and audit monitoring;*
- *consider possibility of establishing hub and spoke service delivery*

This submission supports these steps. So far, we have not heard of any developments to do that with RAIRDs, despite both Scotland making steps in this direction, and England commissioning specialised services according to specifications since 2013. For this reason, we do not believe the Welsh Government will make any progress on this unless it commits to the above steps, specifically for rare autoimmune rheumatic diseases, within this MSK framework.

We note action 1 under part 4:

Health Boards should ensure co-produced integrated local care pathways and service provision for people with inflammatory arthritis and rare diseases are in place to

facilitate appropriate and speedy referrals to specialist services and that local services meet relevant NICE guidelines.

This is not included in the list of recommendations on page 9.

We also note that recommendation 22 refers to “specialist rheumatology service”, seemingly when it is referring to a local rheumatology department, or secondary centre.

Elsewhere in this document (page 36) this form of words is used to describe a tertiary centre where patients are referred for more specialist care than a local rheumatology centre can provide.

We are disappointed that, given the number of specific actions for people with inflammatory arthritis, there are no specific actions which will improve outcomes or the organisation of care for people with rare autoimmune rheumatic diseases. We recognise the difficulty of delivering rare disease care to the same standard as these more common conditions, but given the severity of rare autoimmune rheumatic diseases, health boards and the Welsh government should be aiming to give patients with these diseases’ parity.

Alongside the fact these diseases are life-altering and potentially life-threatening, we think comments like these from our 2021 survey show the need for specific actions to support people with rare autoimmune rheumatic diseases in Wales:

“In 2019 I was diagnosed and treated for cancer, and the difference in the care, especially access to a specialist nurse, was so amazing, I can only imagine how much easier it would be to cope with vasculitis if I had something similar, this difference is really upsetting.”

“I lost confidence in the care here in Wales after being unable to see a specialist for months on end while I was really ill, and then being misdiagnosed and told I had to wait even longer for a skin specialist (I did not need a skin specialist, I needed a kidney specialist). That episode led to some permanent damage.”

“Patients with chronic ill health are being failed in Wales. There is no support, no monitoring, no bloods taken, no medication. I worked hard in my career. For the past eleven years I have become incapacitated by my illness. I have lost my career and am now unable to afford private care.”

“I feel the Welsh NHS has abandoned me, I’m fortunate I can pay to access treatment and consultation in England, but there is a desperate need for a centre in Wales to treat patients like myself with specialist care.”

“Rarely get to see anyone. Not unusual for 2-3 years between appointments.”

“I’m often waiting for months despite appealing for help.”

“My diagnosis 12 years ago took so long and was traumatic, I became so much worse than I needed to be before diagnosis and treatment, this fear stays with me.”

“I have felt totally abandoned for the last 5 years since being diagnosed.”

“Having to fight the health board for minimal contact.”

“I was under the care of a health board outside of where I lived initially and as it was a specialist centre I had very good care. However, I've been repatriated to my local health board and since then the care I've had has been awful, in fact it's been non-existent.”

Comments like these did come alongside others which praised individual healthcare professionals and acknowledged the pressures they work under. However, we need action to effect a system-change which means all patients are supported and all healthcare professionals are able to deliver the excellent care they want to.

We very much hope more specific actions for rare diseases will be adopted in the final framework, particularly with reference to the commissioning of specialised centres and networks.

Prevention, self-management and independence

Question 6: Does the guidance demonstrate the most effective and efficient approaches to develop self-management skills and maximise independence?

The model for access and condition management of adult long-term musculoskeletal conditions, excluding osteoporosis

Question 7: Does the chapter provide a clear and comprehensive model for healthcare professionals to follow? Are there any further changes required to this model?

Diagnosis, treatment and long-term management of conditions

Question 8: Does the guidance capture all the elements of inflammatory diseases? What effective help looks like? What else can be recommended?

Part four covers inflammatory conditions in general, but its introduction then focuses on inflammatory arthropathies. We recognise that there is a need to respect the fact that inflammatory arthritis is more common, and so more patients accessing services will have these conditions, but other inflammatory conditions, such as RAIRDs, should be highlighted here on page 5 to make clear this section refers to them as well. They affect internal organs and glands to a greater extent than other forms of inflammatory condition. Whilst prognosis varies, they have significant impacts on quality of life and life expectancy.

We agree with many of the aspects of effective care highlighted in this framework for inflammatory conditions and agree with many of the priorities and the vision, but without a more detailed implementation plan, systems to measure progress, and hold health boards accountable, the framework will not achieve the specific actions listed. Fundamentally, the vision presented is

a far cry from the care the majority of people with RAIRDs receive currently. Given the reality RAIRDA's surveys seems to show, we think the framework needs to give greater clarity on how exactly these changes will be brought about.

We think the best way to bring the vision for inflammatory conditions into being for RAIRDs is through the commissioning of a specialised centre in Wales, which supports care at local centres through a network.

Additionally, we do not believe the actions or models of care identified in this framework can be achieved, or brought even slightly into being, without addressing the workforce crisis in rheumatology.

Sections on AICTDs, systemic vasculitis, and rare autoimmune rheumatic diseases need to be rearranged

We would recommend amending the sections on rare autoimmune rheumatic diseases, AICTDs and systemic vasculitis so that it is clear that 'rare autoimmune rheumatic diseases' is an umbrella term for AICTDs and systemic vasculitis.

Rather than using facts relating to rare diseases in general, we would rather that the section on rare autoimmune rheumatic diseases references specific information relating to these diseases.

For example:

- Unlike most rare diseases, these diseases lack a simple genetic link. They generally onset in adulthood and predominantly affect women.
- 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.
- 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, nerves, kidneys, heart and glands) and therefore often require similar cross-specialty medical expertise.

Specialised rheumatology services, or service, should be formally commissioned in Wales

We are confused by the mentions of specialised services under the heading of AICTD and systemic vasculitis.

Particularly the sentence: "The budget and accountability for commissioning SRS care is currently a responsibility of the Welsh Health Specialised Services Committee (WHSSC)"

From our understanding, the Welsh Health Specialised Services Commissioning Committee, does not commission any specialised rheumatology services, except from paediatric rheumatology services. From our understanding, tertiary or specialised centres which do exist in Wales are not formally commissioned as such, instead individual clinicians or departments are recognised as having additional expertise in a certain condition, so some clinicians will refer

patients onto them. Formally commissioned specialised services do exist in England, which some Welsh patients do access.

We would absolutely support the specialised commissioning of such services within Wales, alongside access to specialised services in England, we believe this will support the delivery of better care for people with rare autoimmune rheumatic diseases.

The framework should be clearer when referring to specialised rheumatology services and whether it is referring to existing specialised centres in England or specialised services which will be established in Wales, or already exist and will be commissioned by WHSSC in future.

It is important that specialised services are clearly designated and defined, so this clarification is urgently needed. In England, the Clinical Reference Group has agreed guidelines for specialised services since 2013, which means these services have particular aspects others do not. For example, to qualify as a specialised service for rare autoimmune rheumatic diseases and vasculitis, a service needs access to additional support such as: a nephrology service with appropriate expertise, a respiratory service able to manage interstitial lung disease, a day case unit able to provide biologic and cytotoxic infusions, and various other diagnostic tests. It is not just a unit with rheumatologists who specialise in these diseases.

Either using these specifications to develop services in Wales, or developing Wales-specific specifications, is the best way of ensuring specialised centres can deliver good care. The current system of informal recognition is not working.

Access to specialised services must be improved and specialised clinical networks can support this

We note that the framework states that many people with connective tissue disease can be cared for locally. We would argue that it may well be that the issues with local rheumatology services outlined above mean that some departments are unable to deliver care to a ‘national standard’ as the framework says they should.

The framework, in the section on AICTDs, says that “SRSs coordinate care for those who have complex or severe AICTDs on a network basis. These networks are centred on a few hubs that link with other regional rheumatology service providers.”

Networks can support the care of all rare autoimmune rheumatic diseases, not just connective tissue diseases.

We would note that other nations in the UK, particularly England, have recognised that not all rheumatologists have the same knowledge, experience, and confidence in treating and caring for people with these diseases, or access to the technical and organisational resources to allow them to deliver quality care. It is for this reason that England has commissioned specialised centres for these diseases and that Scotland is working to establish a National Managed Network for Vasculitis.

We absolutely support the principle of networks, but they need to be properly established.

Specialist centres, and the clinicians with a particular interest in these conditions who lead them, will have more experience and knowledge of rare autoimmune rheumatic diseases than clinicians

in local centres. In part, because of the organisational and technical features commissioned specialist centres can have access to (if properly commissioned), but also because clinicians in these centres can clearly accumulate experience in caring for these conditions that clinicians in smaller departments, with fewer rare disease patients, will not.

The following principles, laid out in the specialised rheumatology specification in England, should be adopted in Wales:

For some patients, general rheumatologists may only require advice from a distance on diagnosis or management, and the specialist centre will provide telephone helplines and email correspondence to facilitate this. For other patients, they may need to be seen once in the specialist centre, a management strategy instituted, and then followed up by their general rheumatology service thereafter, with rapid access to the specialist team if necessary.

This wording is far clearer than the current wording in the framework. Currently pages 36-38 say that “the majority of people with rare rheumatological diseases in Wales receive most of their care at their local hospitals” and that “Milder manifestations of all AICTDs can be diagnosed and managed in all local rheumatology units”. The former ignores the capability of specialised centres to *support* local care, and the latter ignores the reality that AICTDs can fluctuate and flare, requiring more specialised support at certain times.

Given the issues we note elsewhere with capacity in rheumatology units. We do not think it is accurate to say “all” local rheumatology units can care for these conditions. We also think this attitude would unnecessarily discourage units seeking advice and support from specialised services.

We are concerned about the lack of access to specialist care in Wales. Other UK nations acknowledge the need to invest in specialised services for these patients, which goes above what all local rheumatology departments can offer. The individual patient funding request (IPFR) system is far too restrictive and does not support the kind of care outlined above. We agree with the acknowledgement in this framework that networks would be far more ideal.

In our survey of patients in Wales in 2021, we found that 80% of patients had ever accessed care for their condition at their local hospital’s rheumatology department. Only 11% reported accessing care at a more specialised centre in Wales which they’d be referred onto by another doctor. 29% reported accessing care in a specialised centre in England, although comments indicate that many did so privately.

We also asked patients if they had ever asked for a referral to care to another health board. In total 29% had asked for such a referral, with roughly half having such a referral accepted and half having a referral denied.

Whilst we did not ask patients specifically about accessing private health care in our 2021 survey of patients living in Wales, some commented that they had and many reported that this had been an extremely difficult choice for them to take, or that they could only afford to access this care for a limited time. In 2018, we asked all patients whether they had accessed care privately. In Wales, 38% of patients said they had compared to 28% in the rest of the UK. We think this is further evidence that there is a need for easier access to specialist care for these conditions in Wales.

On top of this, in 2018, only 27% of patients said they had a coordinated plan in place for their care, compared to 40% across the rest of the UK. This further shows the need to commission specialised centres and networks.

We believe that our survey of Welsh patients in 2021 shows that patients are very much prepared to travel further, as the establishment of a specialised service or services would imply. We asked patients how far they would be willing or able to travel to access care at a more specialised centre for their condition. In response, 80% they would be willing to travel further, with 46% saying they would be willing to travel 2 hours more or longer. Only 2% said they would not travel any further, 9% said they already accessed care at the most specialised centre they could. 9% were unsure.

Patients have also told us that it is not necessarily the *distance* travelled, but the *amount* of travelling. For example, patients would far rather travel once, further, to a specialist centre which actually resolves their issues, rather than having to make repeated visits to their GP or local hospital which takes more visits and leaves an issue potentially unresolved.

Mention of networks for rare diseases needs expansion

As noted above, the section on ‘Rare Autoimmune Rheumatic Diseases’ and AICTDs makes mention of the use of networks to support specialised care. However, these networks do need proper support and implementation,

The draft embargoed Getting It Right First Time (GIRFT) national specialty report for Rheumatology services in England notes there is a huge variation across regions in the proportion of departments that are actively engaged in specialised rheumatology networks within each region of England. We would recommend that health boards and the WHSSC learn the lessons from England’s experience.

Part of the issue has been the lack of firm guidance in what these networks need to be effective. The draft embargoed GIRFT report suggests a number of principles which seem crucial for an effective clinical network including:

- Clearly defined network roles, including a coordinator and clinical lead.
- Job planning for these network roles.
- Networks should aim to use data to drive service improvement.

We would note the importance of job planning: these networks clearly need some investment of resource in order for them to be established. We believe that the investment required in such networks is worthwhile as it enables more consistent standards of care across the region, which will reduce the number of patients developing complications and organ damage which are clearly costly to the health service.

We welcome that this guidance seems to propose moving away from a model of individual patient funding requests (IPFRs) and towards the use of networks.

Patients believe that IPFRs are a major barrier to accessing the best care for their conditions. Clinical networks where specialist centres support local hospitals to deliver care will mean more patients get better care locally, and could offer a more collaborative approach to care.

However, we would note that these networks do not mean that patients do not need to travel to specialised centres, this will still need to occur and not all care can be delivered locally, even with specialist support.

We would welcome urgent engagement with the Welsh Government to understand how a specialised network and specialised centre (or centres) will be commissioned and supported.

Care in England is not mentioned at all

Currently, patients in need of care from a specialist centre, in line with the detailed expectations of such a centre in the service specification laid out above, or in need of attention of a specialist in an individual condition, may be referred, or ask to be referred to a service in England.

Even if Wales does establish a specialist rheumatology service, some patients will still need referral to England. This would be in line with practice in England, where specialist centres exist in each region, but some patients are still referred out of their region to see nationally recognised specialists.

29% of patients have had some care from a specialised centre in England whilst living in Wales, although between a third and a half of those patients have done so privately. The small number (just under 10%) of patients who accessed care in specialist centres in England and also had their care coordinated by these centres, had a higher confidence in the services that cared for them, higher confidence in their doctors, and higher confidence in their nurses.

However, it remains extremely difficult for patients to access care in England, despite them, and sometimes their doctors, feeling they would clearly benefit. Some patients report that, doctors, including GPs and specialists, have to fight or engage in collusion to allow patients to continue to get care in England. When both patients and clinicians agree that patients cannot get the care they need without bending or breaking the rules of the system, something is clearly wrong.

Some patients did report issues with their care when it was shared across national borders, but patients also reported issues across the borders of Welsh health boards.

“Needs to be better coordination between tests done at Welsh hospitals and the English specialist hospital and vice versa. Currently the English specialist can’t see my Welsh bloods and it means I have to have repeats which is just a waste of time and money”

“As my care is split between my rheumatologist (based in England) and my GP in Wales, and a neurologist in Wales, there are problems communicating results and care between Wales NHS and England NHS. This affects my treatment and care.”

“Records not being transferred between health boards effectively or efficiently which has caused a delay in receiving the correct medication.”

We recognise that this framework is centred on care delivered in Wales, by Welsh health boards, but when the care for these conditions is shared with services in England, it is disappointing that

these are not mentioned, particularly when there are issues with referrals and coordination to these specialist centres.

Multi-speciality care and clinics are rare in Wales

The importance of multidisciplinary team input, and for well-coordinated care across different specialties is noted for inflammatory conditions. For rare autoimmune rheumatic diseases (RAIRDs), which may involve multiple organ systems, this is particularly important.

In RAIRDA's 2018 survey, only 9% of patients living in Wales reported they had ever accessed care at a clinic with doctors from multiple different specialties. This compared to a, still low, 17% across the rest of the UK. This is despite 93% of UK survey respondents reporting seeing clinicians from multiple specialties.

The proportion of patients who told us in 2018 that they had a coordinated care plan in place was low across the UK. However, it is substantially lower in Wales at 27% compared to 40% across the UK.

Formally commissioning a specialised centre or centres for RAIRDs which has/have access to specific specialties, as occurs in England, will at least support better cross-speciality access in these centres.

Patients commented that the multi-system nature of these conditions was not appreciated by healthcare professionals – again, ensuring better cross-speciality care and specialist expertise is important. This should make sure this is not just understood, but that care is delivered in a way which respects this.

Good mental health is supported by good care

In 2020, our survey of patients across the UK found issues with access to psychological support across the UK. However, patients in Wales were less likely to say they had been offered psychological support, with under 10% saying they'd been offered it.

Access to this support should be improved, but it should also be acknowledged how negative interactions and poor communication can impact the mental health of someone with these conditions. This should be acknowledged throughout this MSK framework.

Poor communication and coordination of the care for someone's physical condition (or 'negative interactions') do have substantial negative impact on mental wellbeing. This has been shown by research but also through patient comments in our own surveys ("Is it me? The impact of patient-physician interactions on lupus patients' psychological well-being, cognition and health-care-seeking behaviour" by Sloan et al.)

We know that patients often raise issues with nurses that they feel unable, or feel they have insufficient time, to talk with consultants about. Such as the impact of their conditions on their personal, family or working life. Access to nurses is important for this reason, but also because they perform a role providing communication, coordination, and responsive care.

"Greater understanding of the emotional toll these conditions put on patients could be instilled, it's taken a breakdown to get somewhere with treatment."

“The care sometimes feels very disjointed and not very communicative. It feels very lonely to have an autoimmune condition in Wales.”

“Records not being transferred between health boards effectively or efficiently which has caused a delay in receiving the correct medication. This has also impacted on my health and mental wellbeing.”

“My mental health has been worn down by my vasculitis journey, I think mainly through feeling alone when so extremely ill.”

“Mental health is not being treated alongside having a lifelong debilitating illness. Since my renal failure I asked to see someone for my low mood. For a year now I am waiting for a referral as the waiting lists are massive!”

Care delivered by primary care and shared care can work well, but it needs support from secondary/tertiary care

One point that should be emphasised is the fact that primary care services, and particularly GPs, are often ill-equipped to support many people with these rare diseases. Many GP clinics will see very few patients with any of these conditions over the course of their entire careers, and each clinic most likely only has a single or a couple of patients with these diseases. This means even the care GPs can give, can only be given by GPs who have heavy involvement and strong relationships with secondary care, and that GPs struggle to recognise the diffuse and complex symptoms of these rare disease.

For continuing care, this does mean that hospitals need to be responsive to their patients’ needs, particularly flares or adverse reactions to medication. Without this support, patients report feeling lost, as their GP is unable to offer much support at all.

In our 2018 survey, 62% of patients in Wales said they were “not at all” or “not very” confident in their GP’s understanding of their condition. In the rest of the UK this was lower at 53%. We are genuinely unsure why this is, given we do not believe there are any substantial differences in GPs’ knowledge of these conditions between the rest of the UK and Wales. We suspect the issue is that hospital departments in Wales are able either to give less support to patients, leading to patients being reliant on GPs for more of their needs (so their lack of confidence dealing with these conditions is more apparent), or that hospitals give less support to GPs to deal with these conditions.

For example, patient comments illustrated this, saying:

“The shared care system usually works, but it depends on rheumatology getting back to my GP. Currently been off medication for a week while GP awaits response from rheumatology which is not forthcoming. Much longer and a flare is inevitable so I will restart the meds without their input.”

Diagnosis times can be most simply shortened by cutting waiting times

In terms of diagnosis, we believe that ‘advice and guidance’ procedures which allow hospital consultants to advise referring clinicians and understanding the reasons for referral earlier and

therefore prioritise patients. This should prevent a situation where a patient who needs to be seen urgently, is made to wait longer, and the consultant only realises the severity of the symptoms they were referred for when it is time for their appointment.

We welcome the mention that referrals will be made from other departments and specialties to rheumatology. We do think this point should be made more explicitly with regard to multi-system autoimmune diseases, not just inflammatory arthritis. Often, a GP may only respond and refer based on symptoms in isolation. For example, a patient with lung issues from undiagnosed scleroderma, may be seen by a respiratory clinician, before other symptoms suggestive of potential autoimmune disease are picked up and a referral is made to rheumatology.

We were concerned that the proportion of patients in Wales waiting longer than 6 months to see a specialist in Wales was 35% compared to 24% in the rest of the UK. Whilst many of the delays involved in the diagnosis of these conditions are difficult to cut, as they are linked to their complex multi-system nature, rarity and ability to ‘masquerade’, we do believe that cutting waiting times would have a significant impact in cutting the total wait for diagnosis that people with these diseases face.

All patients should have access to advice lines and email inboxes to raise issues at short notice

The use of nurse-staffed advice lines or email inboxes in rheumatology services is common across the UK and NICE quality standards for arthritis note that services should give advice to people with these diseases within 1 working day.

People with rare autoimmune rheumatic diseases should be able to expect the same care. These diseases are at least as severe as inflammatory arthritis and many of the treatments (and therefore side-effects) are the same or similar. Although the impact of flares and side effects can also be far more serious.

In Wales, 35% of patients said they did not have the ability to raise issues between appointments, such as disease flares and medication side effects. 82% of patients who could raise such issues did so through calling a specific nurse or a department telephone number with 20% doing so via email. A small number of patients said they called or emailed their consultant’s secretaries directly.

Only 7% of patients have never had to use such methods to raise issues, indicating these services, where available, are well-used and needed. 56% said advice they received when they raised issues was generally prompt and helpful, with 36% saying that responses were often “too slow or staff responding are not confident in supporting people with my rare condition.”

We would recommend that departments and health boards that care for people with these rheumatic diseases make sure their advicelines and email inboxes cater to these patients’ needs as well as those of people with inflammatory arthritis.

“Our rheumatology department run a specialised lupus clinic, and we have access to lupus helpline for advice”

One issue we find across the UK is that nurses may not have confidence in dealing with queries relating to RAIRDs or are unable to escalate issues. Upskilling nurses and making sure they have the support they need is important.

Question 9: Will the actions within this chapter, taken together with the chapters elsewhere in this document improve awareness of rare diseases

The action in this chapter will not meaningfully improve outcomes for people with rare autoimmune rheumatic diseases and we doubt their capability to meaningfully improve awareness.

‘Awareness’ of rare diseases is difficult to achieve, for example, GPs cannot all have a meaningful understanding of all these diseases and increasing a general ‘awareness’ of these diseases may not necessarily support them to make an earlier referral for a suspected rare autoimmune rheumatic disease, or care for their patients with these diseases better, given their variety and complexity.

What would be better is a system where hospitals are more able to give GPs the specific support they need, when they need it.

Within the profession, rheumatology consultants are all ‘aware’ of these rare diseases and will have had to demonstrate an understanding to qualify, but not all have the same level of experience in caring for patients with these diseases.

The best way to increase understanding of these diseases in GPs and other clinicians is through supporting rheumatology departments, and other hospital departments, and ensuring they are not over capacity.

Given rare autoimmune rheumatic diseases are a specific group of diseases with similar clinical characteristics, we do think actions should be more specific to them, rather than rare diseases in general.

Action 1:

We are glad to see rare diseases mentioned on par with inflammatory arthritis here, but we doubt whether this action will actually result in any health boards changing their procedures or pathways. ‘Rare diseases’ is far too broad a term.

Action 2:

The Rare Disease Strategy is now being supplanted by the Rare Disease Framework.

Our key criticisms of the, now obsolete, Rare Disease Strategy were:

- Rare autoimmune rheumatic diseases have no simple genetic link. This means that significant amounts of the strategy, referring to genomic testing are irrelevant for non-genetic rare autoimmune rheumatic diseases.

- The lack of measurable outcomes has meant it is impossible to say whether the strategy, or national action plans, have improved outcomes for people with rare diseases, particularly for those with no simple genetic link.

The UK-wide Rare Disease Framework, which all nations must develop action plans to meet, makes a welcome change in making explicit the need to improve outcomes for people with non-genetic rare diseases.

Given the significant number of patients with rare autoimmune rheumatic diseases, and the clear direction from the Rare Disease Framework to tackle issues relating to non-genetic rare diseases, the Welsh Government's rare disease action plan must make clear how it will improve outcomes for people with non-genetic rare diseases such as rare autoimmune rheumatic diseases. This MSK Framework must make reference to the new Rare Disease Framework, rather than the now obsolete Rare Disease Strategy.

We would welcome a specific recommendation in this section related to the establishment of networks and the move away from Individualised Patient Funding Requests. Such as:

“All rheumatology departments in every health board which care for people with rare autoimmune rheumatic diseases in any capacity, must be part of a specialised rheumatology network and move away from the use of Individual Patient Funding Requests.”

This is a specific action which all health boards can take to support people with these rare diseases. Directing departments, hospitals and health boards to join these networks should be done alongside issuing clarification regarding the situation with the commissioning of specialised centres for these conditions, making clear whether WHSSC is now going to take this on.

As noted in our response to question 8, Wales must learn from the experience of England in establishing these networks to ensure they achieve their objectives.

Without a specific action, relevant to rare diseases covered by this MSK Framework, we do not believe this framework will have any positive impact on the awareness of rare diseases or the outcomes from rare diseases.

Workforce development, education and recruitment

Question 11: How can this chapter be strengthened to address workforce education and recruitment?

It is disappointing that this section only asks health boards to further understand the demands on their rheumatology services, when we know that demand far outstrips the care these services can supply. We would hope that the evidence presented in the British Society for Rheumatology's recent workforce report, alongside evidence from patients, detailed in this response, which shows that shortages of staff are directly reflected in the quality of care patients receive, is enough to convince the Welsh Government that there is now an urgent need for action to support the rheumatology workforce through recruitment, training, and new ways of working.

Measures to increase specialist training places, urgently recruiting from outside Wales, and finding new ways of working must be implemented.

We support the recommendations of the recent report ‘Rheumatology workforce: a crisis in numbers’ by BSR. Whilst there are issues across the UK, BSR’s report contains significant evidence of issues in Wales. Whilst there are issues with the rheumatology workforce across the UK, we do think these issues are particularly acute in Wales.

For example, in 2018, 24% of patients in the rest of the UK waited over 6 months to see a specialist. In Wales the figure was 35%. This comes on top of evidence cited earlier in this report of overstretched services, such as higher proportions of patients seeking private care and higher proportions reporting a lack of a coordinated care plan.

We understand that the small size of some departments in Wales can mean that in the event of a few consultant vacancies, services can end up reliant on locums. This is inadequate and, at the very least, it means that patients do not receive continuity of care between appointments. When combined with the fact patients struggle to get transferred for their care at another health board, accessing good care can become impossible.

Comments we received from patients provide further evidence of the extent of the crisis:

“My rheumatologist left the health board over 2 years ago and I have been left in limbo with care.”

“The rheumatology dept is vastly understaffed, and waiting list for clinics are long. Therefore conditions deteriorate and end up needing more intensive intervention making the rheumatologists job much more difficult and burdening the waiting list more.

“Rheumatology wait in North Wales about 2 years. My GP told me they were asked to refer as few people as possible in January 2020. I meet blood test criteria, but my GP told me to stick with private care.”

“I have not been seen by rheumatology for about two years as they are so busy.”

“My Respiratory Consultant, Neurologist, Orthopaedic Surgeon and Urologist have tried to help me, many have written to Rheumatology asking for me to be seen, as has my GP.”

Question 12: How can we ensure health and social care professionals are aware of how to treat and manage musculoskeletal conditions effectively?

Clinical networks are needed to ensure that those clinicians with the most expertise and experience in caring for rare diseases can support those clinicians who have less. All rheumatologists have an awareness and understanding of these diseases, but their rare nature means some will have more experience and expertise than others. Networks which support clinicians to deliver the best care will support this.

In our recent survey of patients in Wales, we asked patients whether they were generally confident that their rheumatology doctor(s), rheumatology nurse(s) and doctors from other specialties (who had cared for them after their diagnosis) understood their condition.

Overall 52% of those cared for by a rheumatologist said they had confidence in their understanding of their rare autoimmune rheumatic disease. The confidence in rheumatologists was highest among those patients who had their care coordinated by some kind of specialised centre in Wales or England at 73%. We think this is more evidence that there is a need to properly commission a specialised centre, establish networks to support local rheumatologists to care for these patients, and increase the ability for patients to access care across health board boundaries.

55% who had a rheumatology nurse involved in their care were confident in their nurse's understanding of their condition. This demonstrates that despite these diseases' rarity and complexity, nurses can still perform an absolutely crucial, and much valued role supporting patients, but there is a further need to upskill and support nurses to care for these patients.

We think there is compelling evidence to support the proper commissioning of at least one specialised centre for the care of these diseases, along with a network which allows it to support better care elsewhere.

Fewer patients, 38%, had confidence in doctors' from other specialties (such as respiratory, cardiology, nephrology) understanding of their conditions. We think this shows the importance of rheumatology departments to be able to support these clinicians. Overburdened rheumatology departments are likely to struggle to do this.

As noted above, patients' confidence in their GPs understanding of their condition is low, and lower in Wales than the rest of the UK, and this is something which does matter to patients. However, we think the best way to support GPs and change this is better support from hospitals, rather than trying to improve the understanding of every GP of these rare diseases.

Evidenced based care and innovation

Question 13: Is there any recent research, guidance or evidence not included in the document that you are aware of which should be taken into account to better enable people to live with arthritis and musculoskeletal conditions?

RAIRDA's full report on our 2018 survey, 'Reduce Improve Empower' is available [here](#).

Evidence on early diagnosis:

[Impact of early versus late systemic lupus erythematosus diagnosis on clinical and economic outcomes.](#)

Oglesby A, Korves C, Laliberté F, Dennis G, Rao S, Suthoff ED, Wei R, Duh MS. Appl Health Econ Health Policy. 2014 Apr;12(2):179-90. doi: 10.1007/s40258-014-0085-x

Conclusion: Patients diagnosed with SLE sooner may experience lower flare rates, less healthcare utilization, and lower costs from a commercially insured population perspective. This finding needs to be further explored within the context of background SLE disease activity.

[Delayed diagnosis adversely affects outcome in systemic lupus erythematosus: Cross sectional analysis of the LuLa cohort.](#)

Kernder A, Richter JG, Fischer-Betz R, Winkler-Rohlfing B, Brinks R, Aringer M, Schneider M, Chehab G.
Lupus. 2021 Mar;30(3):431-438. doi: 10.1177/0961203320983445. Epub 2021 Jan 5. PMID: 33402036;

Conclusion: In systemic lupus erythematosus, longer time to diagnosis was associated with worse outcome. Concepts in care with the intention to shorten the time to diagnosis are needed to improve the long-term outcome of the disease.

The following articles lay out the challenges in communication between patients and clinicians:

[Things left unsaid: important topics that are not discussed between patients with systemic sclerosis, their carers and their healthcare professionals-a discourse analysis.](#)

Denton CP, Laird B, Moros L, Luna Flores JL.

Clin Rheumatol. 2020 Sep 11. doi: 10.1007/s10067-020-05371-2. Online ahead of print.

PMID: 32915347

Conclusion: Key topics, including mortality and prognosis, are rarely openly discussed, leaving patients uncertain and anxious about the future. By communicating about difficult but important topics, physicians and nurses could help patients and carers manage and plan their lives. This study shows that a multi-professional team-based communication approach is likely to better address patient needs and priorities.

[Is it me? The impact of patient–physician interactions on lupus patients’ psychological well-being, cognition and health-care-seeking behaviour](#), Melanie Sloan, Felix Naughton, Rupert Harwood, Elliott Lever, David D’Cruz, Stephen Sutton, Chanpreet Walia, Paul Howard, Caroline Gordon, Rheumatology Advances in Practice, Volume 4, Issue 2, 2021

Conclusion: Negative medical interactions pre- and post-diagnosis can cause a loss of self-confidence and a loss of confidence and trust in the medical profession. This insecurity can persist even in subsequent positive medical relationships and should be addressed. Key physicians implementing empowering and security-inducing strategies, including being available in times of health crises and validating patient-reported symptoms, might lead to more trusting medical relationships and positive health-care-seeking behaviour.

[The impact of the COVID-19 pandemic on the medical care and health-care behaviour of patients with lupus and other systemic autoimmune diseases: a mixed methods longitudinal study](#), Melanie Sloan, Caroline Gordon, Rupert Harwood, Elliott Lever, Chris Wincup, Michael Bosley, James Brimicombe, Mark Pilling, Stephen Sutton, Lynn Holloway, David D’Cruz Rheumatology Advances in Practice, Volume 5, Issue 1, 2021

Conclusion: The diversion of resources away from chronic disease care was perceived by many participants to have caused adverse outcomes. Fear about increased vulnerability to COVID-19 was high, contributing to health-care-avoidant behaviours. This study also highlights the influence of clinician accessibility and patients feeling medically supported on multiple measures of physical and mental health.

[Medically explained symptoms: a mixed methods study of diagnostic, symptom and support experiences of patients with lupus and related systemic autoimmune diseases](#), Melanie Sloan, Rupert Harwood, Stephen Sutton, David D’Cruz, Paul Howard, Chris Wincup, James Brimicombe, Caroline Gordon, Rheumatology Advances in Practice, Volume 4, Issue 1, 2020,

Conclusion: Patient responses indicated that timely diagnosis could be facilitated if physicians had greater knowledge of lupus/related systemic autoimmune diseases and were more amenable to listening to and believing patient reports of their symptoms. Patient priorities included physicians viewing them

holistically, with more emotional support and assistance in improving quality of life, especially in relation to fatigue.

Additional questions

Question 14: What third sector provision and support can be utilised to support this area?

Action 13 discusses the use of social prescribing to signpost service users to sources of information and community based resources. We have found that the existing social prescribing framework does not provide adequate support for rare diseases. Often, this is because information and support is primarily provided by national, rather than local, charities due to the small number of patients. For rare diseases, social prescribing should expand to national organisations for the benefit of patients.

Some charities supporting RAIRD patients have grant-funding programmes for services such as Lupus Clinical Nurse Specialists.

Question 15: Are there any terms or phrases in the document you feel would benefit from further explanation in a glossary?

Question 16: Certain conditions are known to disproportionately affect people with particular characteristics, such as gender or race. Are there any further considerations required to address any inequity of treatment?

RAIRDS disproportionately affect women. For example: 9 in 10 people with lupus are women and around 8 in 10 people with scleroderma. Sjogren's is also far more common among women.

Lupus is disproportionately prevalent among people from Black, African, Caribbean or Black British and Asian or British Asian groups. Disease also tends to be more severe and these patients have a lower life expectancy.

Question 17: We would like to know your views on the effects this guidance would have on the Welsh language, specifically on opportunities for people to use Welsh and on treating the Welsh language no less favourably than English.

What effects do you think there would be? How could positive effects be increased, or negative effects be mitigated?

Question 18: Please also explain how you believe the proposed Framework could be changed so as to:

- have positive effects or increased positive effects on opportunities for people to use the Welsh language and on treating the Welsh language no less favourably than the English language.
- have no adverse effects on opportunities for people to use the Welsh language and on treating the Welsh language no less favourably than the English language.

Question 19: We have asked a number of specific questions. If you have any related issues which we have not specifically addressed, please use this space to report them:

Please enter here: