A13/S/a

2013/14 NHS STANDARD CONTRACT
FOR SPECIALISED RHEUMATOLOGY SERVICES (ADULT)

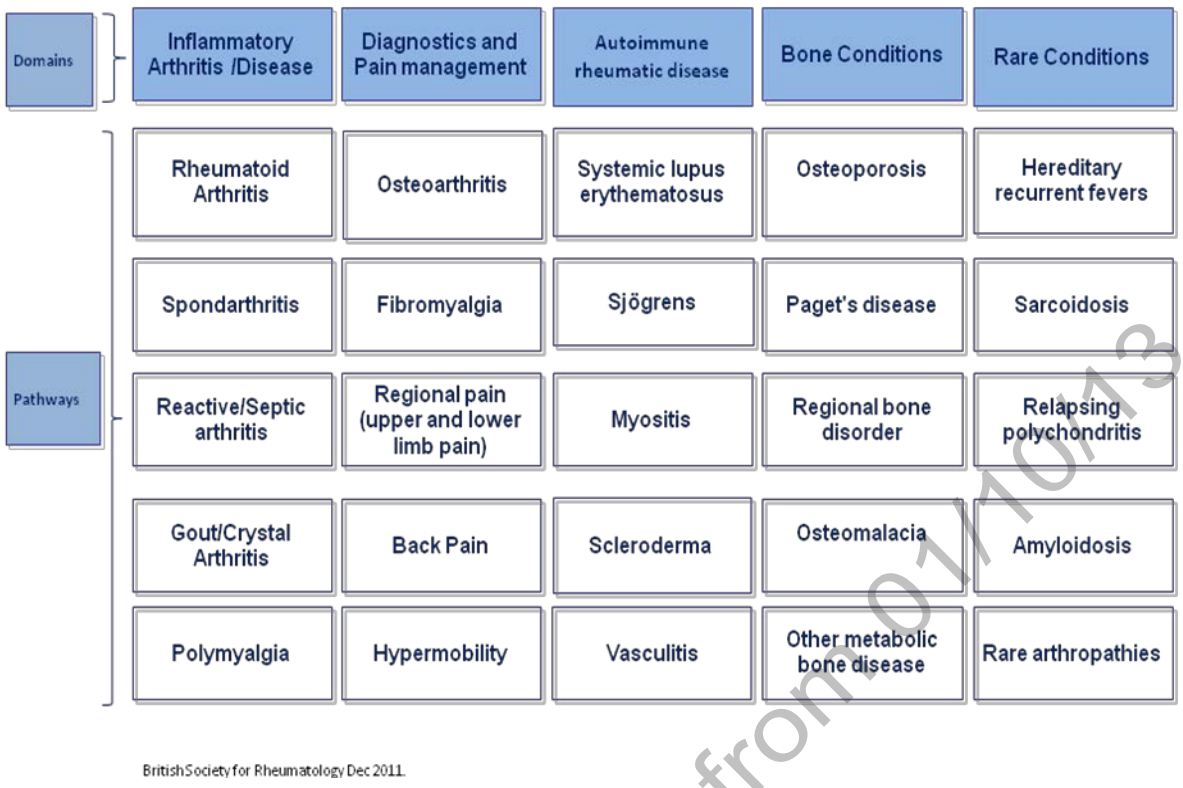
PARTICULARS, SCHEDULE 2- THE SERVICES, A- SERVICE SPECIFICATIONS

Service Specification No.	A13/S/a
Service	Specialised Rheumatology Services (Adult)
Commissioner Lead	
Provider Lead	
Period	12 months
Date of Review	

1. Population Needs

1.1 National/local context and evidence base

Rheumatology is a multidisciplinary speciality and the rheumatologist works in close liaison with other medical specialists and health professionals. Rheumatology deals with the investigation, diagnosis and management of patients with arthritis and other musculoskeletal conditions. This incorporates over 200 disorders affecting joints, bones, muscles and soft tissues, including inflammatory arthritis and other systemic autoimmune disorders, vasculitis, soft tissue conditions, spinal pain and metabolic bone disease. A significant number of musculoskeletal conditions also affect other organ systems. Rheumatological conditions affect all age groups and there is a corresponding service specification for Specialised Paediatric Rheumatology Service. The breadth and depth of rheumatology is illustrated in the diagram of rheumatology domains and pathways (below):



This specification uses two broad categories to classify specialist rheumatological disorders:

A. Inherited Disorders of Connective Tissue
B. Rare Metabolic Sclerosing and Dysplastic Bone Diseases

A. Inherited disorders of connective tissue include:

- Ehlers Danlos Syndrome (excluding type III), which is already nationally commissioned
- Marfan syndrome and related conditions
- Pseudoxanthoma Elasticum
- Arthrogryposis Multiplex Congenita

B. Rare Metabolic, Sclerosing and Dysplastic Bone Diseases. These include:

- Osteogenesis Imperfecta
- Fibrous Dysplasia
- Chondrodysplasias (including Stickler syndrome (diagnostic service nationally commissioned) and related conditions)
- Tumoral Calcinosis
- Fibrodysplasia Ossificans Progressiva
- Fibrogenesis Imperfecta Ossium
- The Osteopetroses and Melorheostosis
- Conditions manifest by metabolic and enzyme defects in bone, e.g.
    - Hypophosphatasia
    - X-linked hypophosphataemia

- Homocystinuria
- Hypoparathyroidism
- Pseudohypoparathyroidism
- Alkaptonuria (nationally commissioned))

These conditions will link with other specialised services including:

- Assessment and provision of equipment for people with complex physical disability (all ages)
- Specialised rehabilitation services for both brain injury and complex disability (adults and paediatric);
- Specialised orthopaedic services (adult and paediatric);
- Specialist neurological/neurosurgical services
- Specialist genetics services (adult and paediatric);
- Specialised musculoskeletal radiology services (adult and paediatric);
- Specialised pathology services (bone pathology and biochemistry);
- Specialised cardiology, ophthalmology services (multisystem diseases of connective tissue and bone can often associated with rare and other eye and cardiac lesions).

Autoimmune Rheumatic Disease and Vasculitis

This category includes the following conditions:

- Autoimmune rheumatic disorders:
  - Systemic lupus erythematosus (SLE)
  - Antiphospholipid syndrome (APS)
  - Systemic sclerosis
  - Sjogrens syndrome
  - Inflammatory muscle disease (myositis)
  - Overlap syndromes
- Vasculitides, including:
  - Antineutrophil cytoplasmic antibody (ANCA) associated vasculitis: Granulomatosis with Polyangiitis (Wegeners), Microscopic Polyangiitis, Eosinophilic Granulomatosis with Polyangiitis (Churg Strauss Syndrome)
  - Polyarteritis nodosa
  - Takayasu's arteritis
  - Henoch Schonlein purpura
  - Cryoglobulinaemia
  - Behçet's syndrome and related disorders (highly specialised service)
  - Rheumatoid vasculitis
  - Giant cell arteritis
  - Hypocomplementaemic urticarial vasculitis
  - Blau syndrome
- Other rare inflammatory disorders:
  - Eosinophilic fasciitis
  - Autoinflammatory Periodic Syndromes

- Relapsing polychondritis

The national context for autoimmune rheumatic diseases is reviewed in “Improving NHS services for rare autoimmune diseases”, a review by an independent expert clinical group on rare autoimmune disorders.

Rare autoimmune diseases affect all ages of the population from Kawasaki disease in young children to giant cell arteritis in the elderly.

In considering rare inherited disorders of connective tissue, rare metabolic, sclerosing and dysplastic bone diseases and rare autoimmune connective tissue and other inflammatory diseases, there are a number of challenges in the diagnosis and treatment:

Professional awareness and ability to spot signs and symptoms

The Behçet’s Syndrome Society found that the average delay for diagnosis for Behçet’s disease is 12 years. Similarly, LUPUS UK has reported that the average length of time their members have had symptoms prior to diagnosis is 7 years. These delays can be fatal with patients experiencing advanced disease, organ failure or even dying before they are diagnosed.

Challenges in diagnosing effectively

Achieving an accurate diagnosis requires a patient being referred to a specialist with appropriate expertise. Access to appropriate investigations and specialist pathology is therefore critical. High quality access will include short waiting times for tests and results, as well as good communication between specialist and radiologist / pathologist to enable appropriate dialogue on possible diagnoses and required follow-up tests.

Access to appropriate specialist support

The treatment of rare conditions, particularly patients with multi-system autoimmune conditions, is complex and requires a multidisciplinary team. The principles of treatment of inherited connective tissue diseases, or rare bone diseases, or autoimmune diseases are the same across specialities but training and experience often reside within one speciality. For example, a patient may have neurological and renal complications associated with their autoimmune disease, as well as systemic issues, but require a therapy widely used in rheumatology. By operating in multidisciplinary teams, colleagues from other specialities are able to be drawn into appointments and patients receive a more sensitive and timely diagnosis. High levels of specialisation, operating at a large population level, are required to make multidisciplinary teams practicable. Otherwise services will not have sufficient throughput to support a comprehensive multidisciplinary team or to develop the requisite level of specialisation. There is good evidence that specialist services which achieve a high volume of care deliver better outcomes.

The aim of specialisation is to reduce the development of disease-related morbidity and mortality, through a commitment to ensure early identification of patients with complex multisystem disease and ensuring that they have timely access to specialist care.

Continuity of care

Due to the complexities and timings of diagnosis and treatment, complications and secondary conditions that can arise as a result of patients' underlying disease, including vascular disease, diabetes and cancer (autoimmune or inflammatory diseases), or osteoarthritis, fragility fracture, musculoskeletal deformity (inherited connective and bone diseases) it is critical for patients that they are able to be transferred easily between consultants and specialities without having to be referred back to their GP. Tertiary referral to sub-specialty clinics is often required for diagnosis, and management by services with appropriate training and expertise improves outcome and patient experience.

There is evidence from other conditions that the effective use of clinical nurse specialists can help reduce costs by minimising the need for inpatient care or emergency admissions.

Guidelines

Department of Health (2006) 'The musculoskeletal services framework; a joint responsibility: doing it differently'

Guide to Osteogenesis Imperfecta. US NIH 2007 (http://www.oif.org/site/DocServer/paediatricians_guide.pdf?docID=7941)

NICE External aortic root support in Marfan Syndrome IPG394

Map of Medicine topics: Temporal arteritis and SLE
http://www.mapofmedicine.com/

http://www.rheumatology.org.uk/includes/documents/cm_docs/2010/m/2_management_of_giant_cell_arteritis.pdf

Hahn BH, McMahon MA, Willkinson A, et al. American College of Rheumatology guidelines for screening, treatment, and management of lupus nephritis. *Arthritis Care Res (Hoboken).* 2012 Jun;**64**(6):797-808. doi: 10.1002/acr.21664.

Ruiz-Irastorza G, Cuadrado MJ, Ruiz-Arruza I, et al. Evidence-based recommendations for the prevention and long-term management of thrombosis in antiphospholipid antibody-positive patients: report of a task force at the 13th International Congress on antiphospholipid antibodies. *Lupus*. 2011 Feb;**20**(2):206-18.

Bertsias GK, Ioannidis JP, Aringer M, et al. EULAR recommendations for the

management of systemic lupus erythematosus with neuropsychiatric manifestations: report of a task force of the EULAR standing committee for clinical affairs. *Ann Rheum Dis*. 2010 Dec;**69**(12):2074-82.

Mosca M, Tani C, Aringer M, et al. European League Against Rheumatism recommendations for monitoring patients with systemic lupus erythematosus in clinical practice and in observational studies.. *Ann Rheum Dis*. 2010 Jul;**69**(7):1269-74

Bertsias G, Ioannidis JP, Boletis J, et al. EULAR recommendations for the management of systemic lupus erythematosus. Report of a Task Force of the EULAR Standing Committee for International Clinical Studies Including Therapeutics. *Ann Rheum Dis* 2008 Feb;**67**(2):195-205.

Kowal-Bielecka O, Landewé R, Avouac J, et al. EULAR recommendations for the treatment of systemic sclerosis: a report from the EULAR Scleroderma Trials and Research group (EUSTAR). *Ann Rheum Dis*, 2009;**68**:620–8.

Note: UK Scleroderma Study Group Guidelines which will be firmed up over the next year- as these are the ones likely to be used by the specialist SSc community in the UK

Hatemi G, Silman A, Bang D, et al. EULAR recommendations for the management of Behçet disease. EULAR Expert Committee. *Ann Rheum Dis*. 2008 Dec; **67**(12):1656-62.

BSR and BHPR guidelines for the management of adults with ANCA associated vasculitis. Lapraik C, Watts R, Bacon P, et al. BSR and BHPR Standards, Guidelines and Audit Working Group. *Rheumatology (Oxford*). 2007 Oct; **46**(10):1615-6.

Mukhtyar C, Guillevin L, Cid MC, et al. EULAR recommendations for the management of primary small and medium vessel vasculitis. European Vasculitis Study Group. *Ann Rheum Dis.* 2009 Mar;**68**(3):310-7.

Mukhtyar C, Guillevin L, Cid MC, et al. EULAR recommendations for the management of large vessel vasculitis. European Vasculitis Study Group. *Ann Rheum Dis*. 2009 Mar;**68**(3):318-23.

2. Scope

2.1 Aims and objectives of service

Aim

To maximise patients function, minimise morbidity and prevent mortality, through

expert medical diagnosis and multidisciplinary assessment, with comprehensive clinically appropriate and evidence based multidisciplinary management, in partnership with the patient's local Rheumatology service whenever possible.

The broad objectives of the service are to facilitate:

- Accurate and timely diagnosis, within the applicable NHS waiting times standards utilising best practice in the assessment of these rare diseases, with protocols to enable rapid access for new and existing patients.
- Delivery of evidence based treatment plans (or best practice treatment in rare disorders where limited evidence exists) to improve treatment outcomes (reduced mortality and co-morbidity) and maximise patient's functional ability.
- Consistent and equitable decision making about use of off licence therapies in refractory or relapsing disease.
- Early identification of patients with complex multisystem disease, ensuring that they have timely access to specialist care.
- Appropriate clinical and laboratory genetics management to characterise genotype in clarifying diagnosis and in screening families and offer support and counselling in regard of heritability (for inherited diseases of connective tissue and rare bone diseases).
- Appropriate shared care arrangements between specialties for the management of co-morbidities directly associated with the patients' rare disease.
- Detailed audit of patient outcomes and experience, shared with colleagues in other centres, enabling the dissemination of best practice and appropriate benchmarking of quality. For example, it is proposed that this activity would form the basis of a national registry for some rare autoimmune diseases. The European Skeletal Dysplasia Network provides a useful framework for many rare types of bone dysplasia.
- Integration of patient care between regional / national specialised centres and local services through the use of standardised shared-care protocols, ensuring that support is delivered as close to patients' homes as possible, but access to specialist services is maintained.
- Increased awareness of best practice in the diagnosis and management of these rare conditions through active engagement and shared care with local providers.

2.2 Service description/care pathway

Specialised Rheumatology services for adults are pre-dominantly outpatient based, with a smaller day-case component but will also require access to inpatient beds, up to and including intensive care. They require extensive input from a broad multidisciplinary team (MDT) covering medical, nursing and therapy specialities plus specialised diagnostics and access to specialised orthopaedics in some cases. Access to specialist rheumatological advice should be available outside normal working hours via an on call service.

The core MDT for a specialised Rheumatology Centre is:

- Specialised Consultant Rheumatologist (s)
- Specialist Nurse

- Physiotherapist with specialist knowledge of rare rheumatological disorders
- Occupational Therapist with specialist knowledge of rare rheumatological disorders
- Pharmacist with expertise in immunosuppressive medication
- Rehabilitation Therapist
- Dietician
- Clinical Psychologist / Psychiatrist
- Orthotist
- Specialist Musculoskeletal Radiology (inherited connective tissue and bone);
- Specialised Pathology Services (especially bone, renal, muscle, biochemistry, endocrinology)

To be considered a specialised Rheumatology Centre for Inherited Disorders of Connective Tissue or Rare Metabolic Bone Diseases, the MDT would have to have access to a consultant geneticist with an appropriate subspecialist interest, specialised orthopaedic/cardiovascular/neurosurgical services with a core experience in operating on skeletal manifestations of the rare condition, and endocrinology services.

To be considered a specialised Rheumatology Centre for Autoimmune Rheumatic Disease and Vasculitis, the MDT would require appropriate leadership and administrative support with the following additional members. This service description needs to be linked with service descriptions from other specialties with a major interest in multi-system inflammatory disease:

- A Nephrology service with appropriate sub-specialty expertise, access to a high-volume biopsy service, and the ability to provide Plasma Exchange and Dialysis.
- A Respiratory service with expertise in the assessment and management of interstitial lung disease (ILD). The service would need to be able to differentiate between Interstitial Pneumonias due to either the autoimmune disease, or the drug treatment (disease modifying anti-rheumatic drugs (DMARDs) and Biologics), infectious mimics of ILD and also other ILDs that are unrelated to the autoimmune disease (e.g. Hypersensitivity Pneumonitis). This would require access to an ILD specialist physician, radiologist and a pathology service with the ability to analyse bronchoalveolar lavage (BAL) cell differentials. This model would be appropriate for all autoimmune diseases (Scleroderma, Myositis, Sjogren's syndrome, SLE and Vasculitis) due to the range of ILD phenotypes and related problems in all of these diseases.
- An Otorhinolaryngology service with expertise in nasendoscopy assessment of vasculitis in the upper aerodigestive tract, management of airway stenosis and reconstructive tracheal/nasal surgery.
- A neurology service with expertise in the diagnosis and management of inflammatory neurological disease and myopathy.
- An intensive care unit with the ability to perform assited ventilation, cardiac and renal support, and experience in multi-system inflammatory disorders
- In addition access to the following departments: Ophthalmology, materno- feotal medicine, gastroenterology, dermatology, oral medicine/surgery, clinical and laboratory immunology

- Access to an appropriately staffed designated day case unit that can provide Biologic and Cytotoxic infusion facilties. This service should be supported by clear guidelines, protocols, and pathways for patient care, in which are embedded the key principles of Chemotherapy safety, As outlined in the document “Chemotherapy Services in England: Ensuring quality and safety. A report from the National Chemotherapy Advisory Group AUGUST 2009” adopted where appropriate for use in non-cancer chemotherapy.

Specialist Rheumatology centres must have ready access to the following diagnostic investigations:

- Full diagnostic imaging (e.g. X-ray, Ultrasound, echocardiography, computerised tomography (CT), magnetic resonance imaging (MRI), Angiography) including access to image-guided tissue biopsy),
- Respiratory Physiology,
- Nuclear Medicine including dual-energy X-ray absorptiometry (DEXA) and positron emission tomography - computed tomography (PET-CT),
- A Histopathology service skilled in the preparation and interpretation of biopsies from potential organs (particularly renal, head and neck, nerve/muscle, skin and thoracic including broncho-alveolar lavage, skills in the interpretation of vasculitis) and access to a pathologist experienced in analysing bone tissue.Access to an Immunology laboratory in which test assays, result interpretation and external test referral are under the supervision of a Consultant Immunologist,
- Drug level monitoring (ciclosporin, tacrolimus, mycophenolic acid),
- Haematology, specialist coagulation,
- Genetic testing for autoinflammatory disease, inherited conditions of connective tissue and rare bone diseases and clinical geneticist expertise (screening families and counselling).

Centres diagnosing and managing patients with inherited disorders of connective tissue must have ready access to:

- Genetic tests from an accredited UK Genetic Testing Network laboratory
- Specialist Biochemistry appropriate to the investigation of inherited connective tissue and metabolic bone diseases

Management

Specialist centres must work with each other and other acute hospital providers to establish clinical networks for rare inherited connective tissue, rare bone and autoimmune diseases creating a hub and spoke model with the specialist unit at the centre. The objectives of clinical networks for these diseases would be to:

- Facilitate patient choice
- Ensure sufficient patient numbers to support training and experience across the range of specialties
- Provide an opportunity train and retain clinicians in highly specialised areas
- Enable services to pool expertise
- Supporting smooth transfer of care across organisations
- Advise commissioners on appropriate planning, particularly through Joint

Strategic Needs Assessments
- Develop standards, guidelines and care pathways to facilitate assessment of care quality and promote consistency of care
- Reduce unwanted variation in clinical practice by aligning care amongst and between specialised centres through the use of national commissioning policies and the pursuit of best practice
- Facilitate a platform for greater engagement of clinicians and patients with rare diseases with clinical and laboratory research studies.

These clinical networks should serve populations of 3-4 million, considered to be the appropriate level for commissioning most services for rare autoimmune conditions, inherited conditions of connective tissue and rare bone conditions. A similar model has already been successfully implemented in France for rare autoimmune diseases.

Protocols for referral will be readily available allowing speedy access to specialist services.

All patients with severe or refractory autoimmune disease should be registered with a specialist centre and entered into a Registry to allow systematic analysis of patient outcomes. Plans for registries in other conditions (e.g. Marfan Syndrome, Osteogenesis Imperfecta, Fibrous Dysplasia) would be considered and developed. For some patients, general rheumatologists may only require advice from a distance on diagnosis or management, and the specialist centre will provide telephone help-lines 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. For other patients, their condition will be sufficiently complex or severe to require the majority of their care to be delivered at the specialist centre.

Services will:
- Be led by a Consultant Physician (usually a Rheumatologist) with relevant specialist expertise (ideally supported by a Consultant team).
- Have access to multi-disciplinary expertise, ensuring that the patient's care is determined by the most appropriate intervention for them rather than the limits of the expertise in the room when a decision on treatment is made.
- Treat high volumes of patients, ensuring a sufficient level of specialism to deliver the best outcomes.
- Be clustered around centres of expertise, enabling economies of scale.
- Have protocols in place to enable rapid access for new and existing patients.
- Develop shared care arrangements with local services, ensuring that support is delivered as close to a patients' home as possible but that access to specialist expertise is maintained.
- Take steps to audit their outcomes and share these data with colleagues in other centres, enabling the spread of good practice and appropriate benchmarking of quality.
- Usually be engaged in clinical research in the relevant specialist area, including clinical trials.
- Provide training for rheumatologists (and other trainees) in the assessment

and management of these disorders.

For all patients, a detailed history, examination, and inspection of previous hospital records and results of investigation will be undertaken. A multi-organ assessment of disease extent, disease activity and damage will be recorded using validated systems. Based on these findings a management strategy will be formulated by the multi-disciplinary team, with an informed patient being helped to make decisions on the most appropriate course of action. Having established a strategy, ongoing follow-up in the specialist centre, or remotely in the general rheumatology unit, will measure the impact of interventions in terms of quality of life, ability to work and undertake other social roles, disease activity and damage to organs.

With regard to individual or groups of diseases, the following key principles apply that differentiate management from other conditions. The specialist centre would have a responsibility to enhance the care of all patients with a designated 'specialist commissioned' disease in its geographic area/region. This is likely to be evidenced by the inclusion of patients in national registries, regional audits, quality control/dissemination of best practice evidence.

Broadly speaking, there are a number of generic situations for all rare conditions of connective tissue, rare bone and autoimmune disorders, where patient access to a specialist centre would be required, although the threshold is likely to also be affected by the level of “interest” and skill in the non-specialist centre, and local geography. These would include:

- Access to extended diagnostic advice (clinical and laboratory) for newly presenting patients where there is uncertainty regarding the diagnosis.
- Access to multidisciplinary assessment, particularly where there is non-musculoskeletal organ/system related disease.
- Access to specialist management advice for patients who have failed on their initial choice of first-line treatment or conventional management..
- Provision of additional multidisciplinary counselling for special situations e.g.
- Pre-conception and fertility advice and disease heritability issues.
- Provision of specialist day case care for patients who need infusions of Biologic or Cytotoxic drugs where facilities or expertise are not available in their local centre
- Provision of inpatient care for patients with severe or refractory disease who require hospitalisation or additional organ support.
- Access to specialist surgical interventions e.g. Ear, Nose and Throat (ENT), dermatology, orthopedics.
- Access to novel therapies via Clinical Trials.
- Provision of “second opinions” for patients at the request of their General Practitioner (GP).

Wherever possible the specialised Rheumatology service will work in partnership with the patient’s local healthcare provider(s) to establish shared care arrangements that meet the patient’s needs.

Patients will be discharged from specialised Rheumatology services, back to their referring local clinician or in certain circumstances another clinically more

appropriate local clinician (eg if the diagnosis changes and a change of specialty is indicated) when:

- Their Rheumatology specific diagnosis is confirmed and they require no further specialist management, or it becomes apparent that they do not have a Rheumatology specific diagnosis that fits within the scope of this specification
- They cease to require high cost drugs and / or other specialised interventions within the scope of this specification
- Their condition is stable and / or predictable and it is safe and clinically appropriate for them to be cared for by their local non-specialised services.

There are many generic aspects to the assessment and management of rarer autoimmune disorders: similar patterns of organ involvement occur in many (eg glomerulonephritis), immunological tests are important in diagnosis and there is overlap in treatment protocols (for example with drugs such as cyclophosphamide). It therefore seems likely (and is reflected in current practice) that specialist centres will offer care across the spectrum of severe systemic autoimmune disease, although individual physicians within units may focus on specific diseases (and some units may focus on a single disease. This would give the centres a critical mass of expertise and also allow support for an out of hours service. In larger conurbations, the centre may be formed by a consortium of physicians in nearby hospitals. There are, however, more specific aspects of the major rheumatological autoimmune diseases which need to be addressed as part of any specified specialist service. These are outlined below:

Inherited disorders of connective tissue

Marfan Syndrome. Has an incidence of 1:20,000. It is inherited as an autosomal dominant trait and manifest mainly by musculoskeletal, ocular, and cardiovascular abnormalities. Specialist diagnostic input requires: informed specialised clinical and genetic assessments; and management often requires specialist cardiology, pathology and ophthalmological input. Orthopedic input and spinal scoliosis surgery (specialised surgery) is occasionally required.

Stickler Syndrome, PseudoXanthoma Elasticum (PXE) and Arthrogryposis are rare conditions for which no specific drug therapy exists but for which specialised multidisciplinary musculoskeletal team input is crucial. These patients benefit from regular specialised review to orchestrate their holistic management given the extent of their conditions reach into every aspect of their lives because of the distribution of their abnormalities and potential for disability. Specialised education and therapy is key in managing their disease.

Rare Metabolic Sclerosing and Dysplastic Bone Diseases

Sclerosing Bone Disorders consist of osteosclerotic and hyperostotic disorders such as The Osteopetrosis Conditions, Pycnodysostosis, Progressive Diaphyseal Dysplasia, Melorheostosis, Fibrogenesis Imperfecta Ossium, Osteopoikilosis, and other conditions. Bone sclerosis is a prominent part of many different disorders (The list of potential disorders needed to be considered in differential diagnosis is >50

diseases-long and many are very rare; Table 1 Chapter 88 Sclerosing Bone Disorders in Primaer on mineral and bone disorders (MBDs) and disorders of Mineral Metabolism 7th edition). Establishing the cause of high bone mass / bone sclerosis is a highly specialist activity requiring Radiological, Orthopedic and Metabolic Bone Physician specialist input. Detailed imaging and its interpretation and details specialised laboratory bone marker and bone biopsy evaluation is frequently required (specialised radiology and pathology services). However many sclerosing bone disorders are multisystem disorders for which general medical services may be required (conventional imaging and laboratory investigations and involving other medical teams eg oncology, hematology). The disease 'area' is an appropriate area in which to consider a tertiary 'virtual' referral process to a 'Specialised Service Team' as a screening tool, to make referrals more efficient Because bone sclerosis can be a more minor part of a wider condition such patients may benefit from quick triage diagnostic service from a tertiary specialist centre then may be referred back to the secondary referral centre but where bone sclerosis might be the major abnormality (true rare bone disease) the patients would be best served remaining within a specialist unit to facilitate MDT management (therapy, metabolic physician, orthopedics, genetics).

Autoimmune Rheumatic Disorders

SLE (Lupus) and antiphospholipid syndrome

There are approximately 20,000-25,000 of patients with lupus in the UK predominantly women with a peak incidence at the age of 25-30. Its onset however varies from children under 5 to adults over the age of 60.

The prevalence is higher in African-Caribbean, South Asian and Chinese populations compared to European whites, and the disease also tends to be more severe in these ethnic subsets (particularly a higher incidence of renal involvement). While criteria have been developed for the classification of SLE, a number of patients do not fulfil complete criteria but quite reasonably can be diagnosed as SLE. In addition, SLE itself frequently overlaps with other connective tissue disease such as Sjogren's syndrome, antiphospholipid syndrome, systemic sclerosis and myositis.

At the time of diagnosis, many will have symptoms related to their disease for at least 5 years. It is not known how much of this delay is due to the insidious nature of initial symptoms (e.g. arthralgia, fatigue) or diagnostic delay due to lack of awareness in both primary and secondary care. The majority of cases are likely to be referred initially to a local rheumatologist or dermatologist although some will have renal disease as their presenting feature, and renal involvement occurs in up to 40% of cases, and can occur many years after diagnosis, though is usually apparent within 5 years. Because of the potential for Lupus to affect any organ system, sequentially or at the same time, effective management involves accurately assessing both disease activity and damage.

Capturing disease activity and distinguishing it from damage (i.e. permanent change) is important. There are several validated scoring systems for this purpose (British Isles Lupus Assessment Group (BILAG) 2004, SLE Disease Activity Index (SLEDAI),

Systemic Lupus International Collaborative Clinics (SLICC)) although these are not in widespread use outside of specialist centres or clinical trials. The aim of drug therapy is to treat disease activity, according to organ involvement and severity, and prevent disease flares.

Hydroxychloroquine, Azathioprine and Methotrexate are commonly prescribed. However, 10 -15% of patients will continue to have high disease activity despite standard therapy. These patients are likely to need more specialist advice, and require IV Cyclophosphamide, Mycophenolate or access to Biologic Drugs e.g. Rituximab. This drug is currently used effectively by many specialist centres to treat patients with highly active disease; however this is an unlicensed indication and does not have Randomised Controlled Trial evidence. Recently Belimumab has been licensed for use in SLE, but has not received approval from NICE.

It is likely that all patients with Lupus Nephritis would benefit from either assessment or advice from a specialist centre, in addition to those with any manifestation which has not responded to standard immunosupression, and women of child bearing age who need additional pre-pregnancy counselling or fetomaternal obstetric care.

For antiphospholipid syndrome (APS) with or without lupus, access to enhanced screening for cardiovascular and neurological disease is required, with indefinite anti-coagulation. Plasma exchange and intravenous immunoglobulin may be required for the subgroup with severe life-threatening disease known as catastrophic APS.

Within a hub-and-spoke model several types of specialist activity can be identified:

- Anecdotal evidence from specialist centres suggests it is not unusual for the primary diagnosis of SLE to be revised particularly in the context of apparently ‘resistant’ disease. Secondly, a number of patients are often significantly overtreated for mild-moderate clinical manifestations. The converse is also seen where inexperience in handling more difficult patients leads to delays in instituting definitive disease modifying therapies. A specialist centre can therefore provide reassurance on the primary diagnosis (or revision of the primary diagnosis), it can also more accurately define the level of therapeutic intervention required.
- Specific Disease Manifestations: Multidisciplinary care in a specialist centre for the management of specific organ involvement namely nephritis, neuropsychiatric involvement, haematological involvement including clotting-related complications and cardiorespiratory disease.
- Multidisciplinary clinics for:
    - Transitional care for young adults with lupus. This population like other transitioning chronic illnesses have specific issues around growth and development, adherence of medications, sexual development drugs and alcohol issues.
    - Pregnancy counselling and antenatal care. Many aspects of care for patients with lupus and related syndromes require specialist input in pre-pregnancy counselling, care through pregnancy itself and management of postpartum period.
    - Refractory disease assessment: This is important in the management of

the more severe and disease spectrum and requires a consistency of approach from experienced clinicians. Refractory patients would include patients who require:

- Unacceptably high-dose of corticosteroids on a regular basis to control disease (> 10 mg prednisolone daily).
- Failure to be controlled on 2 or more standard immunosuppressive agents.

These are the subsets that often are entered into clinical trials of novel therapeutic agents but also may be eligible for a thorough assessment of their disease and clinical status to confirm the refractory nature of the condition. This group also may require combinations of more novel immunosuppressive agents and or biological drugs.

Linkage to patient support groups at the local and national level. A number of Centres of Excellence already exist across the UK have been designated as such by Lupus UK, the main patient support organisation. Patient input at a local and national level should be promoted by the specialist centres.

Specialist Nurse Services**:**
These will include drug education and counselling as well as focusing on more complex drug regimes and primary prevention of complications and co-morbidities specifically, cardiovascular risk, infection and immunisations, women's health and bone protection.

ANCA Associated Vasculitis (AASV)

Antineutrophil cytoplasmic antibody (ANCA)-associated Vasculitis comprises several related conditions that share overlapping clinical features, characterised by involvement of small and medium sized blood vessels in either single or multiple organs (predominately respiratory tract, renal, skin and nervous systems, although any organ can be involved). The incidence is approximately 1200 new cases per year in England and Wales, and increases with age, with peak onset between 60 and 70 years. Without treatment, the condition is fatal; 2 year survival with treatment is approximately 80%, but varies according to age and renal function at onset. The assessment of ANCA- associated vasculitis is complex, both in determining disease extent, disease activity, and damage. Treatment is tailored to all these parameters, but MDT assessment, particularly the inclusion of ENT, respiratory medicine and nephrology is essential to ensure that treatment decisions are based on accurate organ-specific assessment.

Management of all patients with these conditions is likely to be improved by the integration into routine practice of the use of formal disease activity/damage tools and multidisciplinary working to reflect the diversity of potential organ involvement. Standard induction regimes predominately involve high dose corticosteroids and Cyclophosphamide for 3-6 months (or Methotrexate for milder disease). Maintenance agents include methotrexate, azathioprine, mycophenolate, leflunomide and ciclosporin. Approximately 15% of patients will have refractory disease and not achieve remission with standard treatments, the majority of which

are currently likely to be treated with Rituximab (currently an off-licence indication). The use of Rituximab in vasculitis is currently being scoped by NICE, and is also being studied in ongoing clinical trials.

It is likely that there will be other situations apart from refractory disease where this agent will be used, including as an alternative remission induction agent in situations where there is a relative contraindication to the use of Cyclophosphamide, as a remission maintenance agent where the disease has been characterised by previous relapses on alternative maintenance agents, or where alternative remission maintenance agents have not been tolerated due to toxicity. We consider all patients with AASV should be registered with the specialist centres.

Recent use of PET-CT has identified a a significant number of aortitis and large vessel vasculitis in patients with giant cell arteritis (GCA) and polymyalgia rheumatica (PMR) not responding to steroids. Prolonged corticosteroid therapy is associated with a variety of side effects especially when high dose glucocorticoid therapy is employed. Specialised therapy with agents such as cyclophosphamide and biologics such as tocilizumab are being increasingly used and trialled in these conditions. Specialised imaging such as vascular ultrasound are being increasingly used to diagnose and monitor these conditions.

Idiopathic inflammatory myopathies

These are rare disorders (incidence of approximately 500 new cases per year in England and Wales , prevalence around 50 per million) of which the major adult subtypes are polymyositis, dermatomyositis, myositis as part of an overlap syndrome and inclusion body myositis. They cause progressive muscle weakness but are also associated with pathology in other organs, especially pulmonary fibrosis. The differential diagnosis of myopathy is very broad and requires close liaison with neurology services, and ideally combined clinics with a neurologist with expertise in neuromuscular disease. Access is also required to specialist immunology, neuropathology and neurophysiology. A reasonable evidence base exists for treatment with immunosuppressive drugs, rituximab and immunoglobulin but assessment of response to treatment is difficult and requires specialist multidisciplinary assessment. It is anticipated that all patients with inflammatory muscle disease would be registered with specialist services but that management will usually be on a shared care basis. Inpatient facilities will be requierd for patients with severe weakness, swallowing difficulites and respiratory failure. Assessing outcome in myositis is complex and requires detailed physical assessment of muscle strength (preferably with fixed dynamometry) and experience in muscle MRI scanning. Myositis may be a paraneoplastic disorder, detailed assessment of which should ideally include CT/PET scanning.

Primary Sjogren's syndrome

In Sjogren's syndrome, inflammation of the secretory glands causes dryness of the eyes, mouth, skin and other surfaces. The salivary glands can be chronically swollen. Fatigue and joint pain are common. It is predominantly a disease of

women with male patients being quite rare. It is not fully known how common it is but estimates are about 1 in 200 to 1 in 1000 adult women in the UK. About 10-20% of these will have more severe systemic involvement of the skin, lungs, joints, kidneys or nerves and about 5% develop a B-cell lymphoma of the salivary glands. Diagnosis is by autoantibody testing, multidisciplinary assessment by rheumatology, ophthalmology and oral medicine/surgery and lip gland biopsy. Other specialist involvement includes but is not limited to dermatology, gynaecology, renal medicine, haematology, respirology, ear, nose and throat (ENT). Optimizing treatment requires specialist multidisciplinary input.

A national registry has been established (www.sjogrensregistry.org). Biologic therapies targetting B-cells are currently under investigation.

Systemic sclerosis or scleroderma

These rare disorders are characterised by chronic, progressive fibrotic changes in the skin and other organs. Systemic sclerosis affects about 1 in 10000 people, with an annual incidence of around 1 in 200000. About 80% of patients have limited systemic sclerosis, with milder skin changes but often major vascular disease and a long term risk of pulmoary hypertension. Diffuse systemic sclerosis accounts for about 20% of cases with severe, often rapidly progressive skin disease and severe internal organ involvement , espcially interstitial lung disease. The fibrotic changes are difficult to treat (with poor response to conventional immunosuppression) and the mortality is high- 40% at five years in diffuse disease.

Treatment of the organ based complications may be more successful but good supportive care of the complications of the disorder are crucial.

Some mild cases of limited disease may be suitable for treatment in non-specialist services (though many may benefit from a specialist management plan), but specialist assessment and management is likely to be required for all patients with diffuse disease and those with limited disease and severe vascular problems or pulmonary hypertension.

Unexplained weight loss, muscle weakness or breathlessness should all lead to consideration of specialist aseessment.

Close liaison with specialists in mutiple specialities are required including pulmonary hypertension, nephrology, vascular surgery (ischaemia and calcinosis) and intestinal failure are required for optimal management. Specialist MDT support is crucial.

Entry to clinical trials in important in these unusually difficult to treat disorders. Given the rarity, severity, poor response to treatment and need for specialist MDT services, systemic sclerosis care will sometimes be delivered by services which focus largely on this disease, a possible exception to the model of generic care of autoimmune diseases discussed above.

Other rarer autoimmune disorders and vasculitis

Including Polyarteritis nodosa, Relapsing polychondritis, Cryoglobulinaemic vasculitis, Cogans syndrome, Takayasu's arteritis and other forms of large vessel vasculitis, Eosinophilic fasciitis, and other undifferentiated or unclassifiable presentations of severe connective tissue diseases (CTD) or vasculitis. These disorders are very rare and there is only limited evidence to guide best treatment. Best practice would suggest that similar treatment approaches to those used in severe vasculitis or lupus can guide the treatment of these disorders, and the best outcomes in these disorders seem likely to be found in specialist centres.

Autoinflammatory syndromes

These are rare, newly described conditions in which there is abnormal mulstisystem inflammation, associated with deficiencies in the Innate immune system. They can present to any specialty with organ-specific inflammation, and are often under-recognised. Many individuals with these conditions have identifiable genetic associations, which enable targeting of specific biologic drugs. The majority of patients with these conditions are likely to be under the care of Immunology services, and are included in Specialised Immunology Services (http://www.specialisedservices.nhs.uk/library/26/Specialised_Immunology_Services_all_ages.pdf).

2.3 Population covered

The service outlined in this specification is for patients ordinarily resident in England*; or otherwise the commissioning responsibility of the NHS in England (as defined in *Who Pays?: Establishing the responsible commissioner,* and other Department of Health guidance relating to patients entitled to NHS care or exempt from charges).

* - Note: for the purposes of commissioning health services this EXCLUDES patients who, whilst resident in England are registered with a GP Practice in Wales, but INCLUDES patients resident in Wales who are registered with a GP Practice in England.

The service is accessible to all adult patients with a suspected or known specialist Rheumatological condition regardless of sex, race, or gender**.**

Providers must ensure staff attend mandatory training on equality and diversity and the facilities provided offer appropriate disabled access for patients, family and carers.

When required the providers will use translators and printed information available in appropriate multiple languages. The provider has a duty to co- operate with the commissioner in undertaking Equality Impact Assessments as a requirement of race, gender, sexual orientation, religion and disability equality legislation.

Information for patients regarding Rheumatological diseases is available from the British Society for Rheumatology at the hyperlink below:
http://www.rheumatology.org.uk/Patient_Information/

2.4 Any acceptance and exclusion criteria

Referrals will usually be accepted from general rheumatologists, though exceptionally directly from GPs. The service has a duty to query the content of a referral prior to accepting it if the information provided does not indicate that the patient has a condition that fits into the Specialist Adult Rheumatology Service.

If a service chooses to query a referral it must do so within 48 hours of becoming aware of a senior clinician becoming aware of the content of the referral.

Patients who do not have a rheumatological condition that requires the expertise of a Specialist rheumatological service (as described above) are excluded from this service.

This service specification describes the adult Specialist Rheumatological service and therefore under-18s are excluded and they are covered by the Specialised Paediatric Rheumatology Service specification.

The following are not included in specialised rheumatology;

- all new patients presenting with undiagnosed musculoskeletal symptoms for evaluation and initial management,
- patients transferring from paediatric to adult rheumatology services with well controlled disease;
- rheumatoid arthritis, spondarthritis, reactive arthritis, septic arthritis, crystal arthritis, polymyalgia,
- the majority of bone conditions (e.g. osteoporosis, Paget's disease, regional done disorders, osteomalacia, other metabolic bone diseases),
- all patients with autoimmune rheumatic diseases or rare arthropathies, where:
    - the diagnosis is already established and
    - the manifestations are well controlled by conventional management and
    - the patient and the rheumatologist are satisfied with treatment response and progress of the disease.

2.5 Interdependencies with other services

Although musculoskeletal disease services focus, naturally, on disease affecting joints, muscles and bones, many forms of 'arthritis' involve many organs and systems. Thus the specialist rheumatology teams also work closely with other specialties (often involving joint clinics) including:

- Orthopaedics (including liaison with the skeletal dysplasia network and metabolic bone disease specialists)

- Neurosurgery (particularly for craniocervical work, hydrocephalus and spinal stenosis)
- Cardiac surgery (particularly in Marfan syndrome)
- Maxillofacial surgery
- Vascular and plastic surgery (particularly with regard to systemic sclerosis)
- Nephrology (including access to renal biopsy services)
- Dermatology
- Ophthalmology (for uveitis and other inflammatory eye diseases)
- Neurology (CNS inflammatory disease and muscle disease)
- Cardiology (including assessment of pulmonary hypertension)
- Respiratory medicine (including comprehensive lung function testing)
- Critical Care Services
- Ear, Nose and Throat (for assessment of upper airways for granulomatous and other inflammatory disease)
- Oral medicine/surgery (for xerostomia and ulceration)
- Specialist radiology
- Psychology / Psychiatry.
- Gastroenterology
- Social workers and Social Services
- Wheelchair services

As functional impairment or disability is a common component of Rheumatological disease Specialist centres must liaise with local community health services and social services as appropriate to the individual needs of each patient.

The other key interdependency is between Paediatric and Adult Rheumatology Services. Where clinically required paediatric and adult services must work collaboratively and plan for a seamless transition for the patient between services.

3. Applicable Service Standards

3.1 Applicable national standards e.g. NICE, Royal College

"Improving NHS services for rare autoimmune diseases", a review by an independent expert clinical group on rare autoimmune disorders http://www.mhpc.com/wp-content/uploads/2012/09/Improving-NHS-services-for-rare-autoimmune-diseases.pdf

http://www.mhpc.com/wp-content/uploads/2012/09/Improving-NHS-services-for-rare-autoimmune-diseases.pdf
Department of Health (2006) 'The musculoskeletal services framework; a joint responsibility: doing it differently' Available from Department of Health - www.dh.gov.uk

4. Key Service Outcomes

Performance Indicator	Method of measurement
Mortality	In and out of hospital mortality (including cause of death). Comparison with published survival data. Care is required in interpretation of mortality data as more complex cases referred to specialist centres will be likely to have a high mortality
Remission and relapse	Using recognised disease specific measures of disease activity (eg BILAG for lupus, European Vascular Society (EUVAS) for vasculitis. International Myositis Assessment and Clinical Studies group (IMACS) core data set for myositis
Disease related damage	Using recognised disease-specific damage indices (eg SLICC/American College of Rheumatology (ACR) Damage Index, Vasculitis Damage Index), Rodnan Skin Score plus measures of organ specific damage (eg renal impairment and progression to End Stage Renal Failure, cardiorespiratory assessment).
Functional capacity/ Disability	HAQ or similar, using validated disease specific measures where possible (eg Scleroderma Functional Index)
Comorbidity	Cardiovascular events, osteoporotic fracture, anxiety and depression
Comorbidity prophylaxis	Evidence of screening and preventative strategies (Cardiovascular risk, Fracture risk assessment or similar
Quality of life	EQ5D and disease specific measures if available (eg Lupus Quality of Life)
Patient / carer satisfaction	Questionnaire survey
Access to support groups and education	Questionnaire survey plus patient/ carer participation
Waiting times and numbers	Hospital data systems
Maintenance of Disease Registry	Registry established. Evidence that data is complete/ up to date/ accurate
Participation in clinical trials	National Institute for Health Research (NIHR), Comprehensive Local Research Network (CLRN) and pharmaceutical trials
Evidence of programme of joint working with non-specialist centres	Shared Care Protocols, Outreach Clinics

The data for the items described below can be collected on the UKVAS (UK Vasculitis group) registry.

Referral:

- Reduced time from symptom onset to diagnosis.
- Reduced time to first review my specialist team.
- Reduced proportion of patients with vital organ failure at diagnosis (brain, ear, eye, gut, heart, lung, kidney, peripheral nerve)
- Percentage of patients with diagnosis reviewed by specialist team

Treatment & management:

- Reduced time from diagnosis to start of definitive therapy
- Reduced time from primary therapy failure to second line (especially biologic) therapy
- Reduced major relapse rate (involving a vital organ)
- Reduced severe adverse event rate (defined by hospitalisation)
- Adherence to quality guidelines on clinic frequency, laboratory monitoring and prophyactic strategies
- Measure of access to and qaulity of multidisciplinary opinion (research agenda)

Outcomes:

- Reduced excess one and five year mortality.
- Reduced irreversible vital organ failure, especially end stage renal disease (ESRD).
- Reduced excess one and five year cardiovascular morbidity.

Increased quality of life:

- Part of the research agenda
- Improved patient satisfaction

Reduced health resource utilisation.

- Number of in-patient days/patient
- Number of excess clinic visits (above quality guidelines)