



Body System: Musculoskeletal		
Session Topic: Connective Tissue Disorders		
Educational Format		Faculty Expertise Required
REQUIRED	Interactive Lecture	Expertise in the field of study. Experience teaching in the field of study is desired. Preferred experience with audience response systems (ARS). Utilizing polling questions and engaging the learners in Q&A during the final 15 minutes of the session are required.
OPTIONAL	Problem-Based Learning (PBL)	Expertise teaching highly interactive, small group learning environments. Case-based, with experience developing and teaching case scenarios for simulation labs preferred. Other workshop-oriented designs may be accommodated. A typical PBL room is set for 50-100 participants, with 7-8 each per round table. <u>Please describe your interest and plan for teaching a PBL on your proposal form.</u>
Professional Practice Gap	Learning Objective(s) that will close the gap and meet the need	Outcome Being Measured
<ul style="list-style-type: none"> • Underdiagnosis of Systemic lupus erythematosus (SLE) because the presenting symptoms and signs are often not specific. • Overdiagnosis of SLE because doctors mistakenly use a positive blood test (present in 5% of the healthy population) by itself to make a diagnosis. • Poor adherence to RA clinical guidelines. • Knowledge gaps with regard to screening and diagnosing RA. • Patients often have poor access to specialists and inadequate insurance coverage. • Patients often have poor adherence to prescribed RA medication therapies. • Compliance with prescribed treatment is often a problem, given the use of strong immunosuppressive medications and resulting side effects in young, 	<ol style="list-style-type: none"> 1. Recognize symptoms of connective tissue disorders through astute history and physical exam is vital when a CTD is suspected. 2. Apply appropriate diagnostic strategies to confirm diagnosis, when CTD is suspected. 3. Develop collaborative care plans emphasizing treatment monitoring and adherence to prescribed therapies. 4. Establish standardized processes for possible referral and coordination of care with a rheumatologist. 	Learners will submit written commitment to change statements on the session evaluation, indicating how they plan to implement presented practice recommendations.



<p>reproductive age women who want to partner and have children. This is a relatively unique demographic/therapeutic problem among the rheumatic diseases.</p> <ul style="list-style-type: none"> • Patients with lupus have an increased frequency of related autoimmune problems, such as Sjogren’s syndrome (i.e., dry eyes, dry mouth) and antiphospholipid syndrome (i.e., clotting problems, strokes, fetal loss), that require additional treatments. 		
ACGME Core Competencies Addressed (select all that apply)		
X	Medical Knowledge	Patient Care
X	Interpersonal and Communication Skills	Practice-Based Learning and Improvement
	Professionalism	X Systems-Based Practice
Faculty Instructional Goals		
<p>Faculty play a vital role in assisting the AAFP to achieve its mission by providing high-quality, innovative education for physicians, residents and medical students that will encompass the art, science, evidence and socio-economics of family medicine and to support the pursuit of lifelong learning. By achieving the instructional goals provided, faculty will facilitate the application of new knowledge and skills gained by learners to practice, so that they may optimize care provided to their patients.</p> <ul style="list-style-type: none"> • Provide up to 3 evidence-based recommended practice changes that can be immediately implemented, at the conclusion of the session; including SORT taxonomy & reference citations • Facilitate learner engagement during the session • Address related practice barriers to foster optimal patient management • Provide recommended journal resources and tools, during the session, from the American Family Physician (AFP), Family Practice Management (FPM), and Familydoctor.org patient resources; those listed in the <u>References</u> section below are a good place to start <ul style="list-style-type: none"> ○ Visit http://www.aafp.org/journals for additional resources ○ Visit http://familydoctor.org for patient education and resources • Provide recommendations to identify the major symptoms and risk factors for the connective tissue disorders dermatomyositis, scleroderma and systemic lupus erythematosus, including age, race, family history and gender. • Provide recommendations to differentiate between dermatomyositis, scleroderma, and systemic lupus erythematosus and other conditions that present with similar symptoms; provide an appropriate diagnosis and/or suggest additional testing when necessary. • Provide strategies and resources for counseling patients on treatment regimens to manage symptoms including combinations of immunosuppressant and/or anti-inflammatory medications, diet, lifestyle, and follow-up appointments. 		



- Provide recommendations for managing possible complications associated with connective tissue disorder treatment.
- Provide recommendations for appropriate diagnostic strategies for rheumatoid arthritis to facilitate early diagnosis of the disease.
- Provide recommendations regarding the use of validated outcome measures to monitor disease activity and progression and ultimately optimize therapy for individual patients with RA.
- Provide recommendations and strategies for implementing guidelines and strategies for combination DMARD therapy to facilitate more appropriate DMARD selections for patient therapy.
- Provide strategies and resources for developing collaborative care plans emphasizing treatment monitoring and adherence to prescribed therapies.
- Provide an overview of current clinical guidelines with strategies for implementation and adherence.
- Provide an overview of current pharmacologic and non-pharmacologic treatment options, including evidence-recommendations for maximizing patient outcomes.
- Provide recommendations regarding guidelines for Medicare reimbursement.
- Provide recommendations to maximize office efficiency and guideline adherence to the diagnosis and management of CTDs.
- Provide an overview of newly available treatments, including efficacy, safety, contraindications, and cost/benefit relative to existing treatments.

Needs Assessment:

Connective tissue diseases are rare, but potentially life-threatening diseases. The reported prevalence and incidence of connective tissue disorders are variable, and dependent on differences in study methodology.¹ The prevalence of overlap syndromes, especially mixed connective tissue disease, is unknown, and polymyositis and dermatomyositis are regarded as very rare rheumatic diseases. The definitive diagnosis of MCTD is often complicated by the fact that the overlapping features tend to occur sequentially.² Dermatomyositis (DM) is a rare disease with an incidence of 9.63 per 100,000 (including all subtypes).³ Systemic sclerosis (SSc) is just as rare with annual incidence estimated to be 10 to 20 cases per 1 million persons; whereas the prevalence is four to 253 cases per 1 million persons.⁴ Physicians often misdiagnose systemic lupus erythematosus (SLE) because presenting symptoms and signs are often not specific, and because doctors mistakenly use a positive blood test by itself to make a diagnosis.⁵ Sjogren's syndrome (SS) frequency appears to increase with age, with a prevalence of about 3% in people above an age of 50 yrs., and the female:male ratio is about 9:1.⁶

An estimated 52.5 million adults in the United States reported being told by a doctor that they have some form of arthritis, rheumatoid arthritis, gout, lupus, or fibromyalgia.⁷ By 2030, an estimated 67 million Americans ages 18 years or older are projected to have doctor-diagnosed arthritis.⁸ Arthritis and other rheumatic conditions are the most common cause of disability among U.S. adults and have been for the past 15 years. In 2004, there were an estimated 744,000 hospitalizations with a principal diagnosis of arthritis (3% of all hospitalizations). Overall, 5 million hospitalizations had a principal or secondary diagnosis of arthritis.⁹ There were 78 million ambulatory care visits with a primary diagnosis of arthritis or other rheumatic conditions,



or nearly 5% of all ambulatory care visits that year. Overall, there were 66 million ambulatory care visits with a primary or secondary diagnosis of arthritis or other rheumatic conditions.¹⁰

Data from a recent American Academy of Family Physicians (AAFP) CME Needs Assessment survey indicate that family physicians have statistically significant and meaningful gaps in the medical skill necessary to provide optimal care and management of connective tissue disorders, including arthritis.¹¹ More specifically, CME outcomes data from 2011 AAFP Assembly *Arthritis*, and 2012 and 2015 AAFP FMX (formerly Assembly): *Connective Tissue Disorders* sessions suggest that physicians have knowledge and practice gaps with regard to appropriate screening; recognizing signs that may indicate a need for testing; selecting appropriate diagnostic and laboratory testing; and better awareness of appropriate treatment medications, including joint aspiration for arthritis.¹²⁻¹⁴ Physicians should be able to recognize common physical manifestations of dermatomyositis and scleroderma; be able to select appropriate screening tests; know when to refer to a specialist; and be familiar with common co-morbidities of connective tissue disorders.

One method of diagnosis for autoimmune and connective tissue disorders is a positive antinuclear antibody (ANA) test, meaning antibodies have identified normal, naturally occurring proteins as foreign and dangerous, essentially causing the body to attack itself. A positive ANA test, however, does not necessarily indicate the presence of a connective tissue disorder, especially since certain medications can cause positive ANA results. To avoid misdiagnosis, family physicians should be aware of patients' medication histories and should also consider other risk factors for connective tissue disorders, such as age, race, family history and gender.⁵ Additional diagnostic measures may include muscle and/or skin biopsy, electromyography and measurement of muscle enzymes.⁹

Because many connective tissue disorders are uncommon, family physicians may suspect a diagnosis but be uncertain how to confirm it. Referral to experienced clinicians or medical geneticists can help confirm or exclude a suspected diagnosis. Additionally, once a diagnosis is made family physicians may be able to use laboratory studies to conduct prenatal testing; newborn screening to identify a condition that may become evident later in life; carrier testing to identify adults who may carry a genetic mutation for a disease; and/or predictive testing to spot individuals at risk for developing a genetic connective tissue disorder.⁸

There is no cure for connective tissue disorders, but specific treatment regimens can help patients manage their symptoms. Due to the inflammatory characteristic of these disorders, anti-inflammatory medications are the most common form of treatment for relieving swelling, redness and pain. The exact choice of medications depends upon the type of connective tissue disorder that is present and the overall health of the individual. Family physicians can work with patients to determine the type and amount of anti-inflammatory and immunosuppressant drugs that is right for them. Family physicians can also recommend lifestyle plans to optimize health and reduce symptoms. A well-balanced nutritional plan, regular physical activity and routine testing to monitor the progress of disease are all important treatment options. Family physicians should encourage patients to ensure adequate hydration by drinking plenty of water and fluids, follow a balanced nutrition plan, maintain a healthy body weight, get plenty of rest, and schedule routine follow-up appointments. They should also assist patients with coordination with subspecialists.⁶



Treatment for connective tissue disorders can cause a number of serious complications to arise, including pulmonary hypertension, heart disease, osteoporosis and/or muscle weakness due to corticosteroid use, and pregnancy complications. Family physicians should be aware of these potential complications and discuss the risks with their patients.⁷ Additionally, because pharmacology for connective tissue disorders is ever-changing, the National Institute of Arthritis and Musculoskeletal and Skin Diseases, the lead federal agency for connective tissue research, is conducting studies to gain a more complete understanding of the diseases, including genetic origins of symptoms, disease progression and mutations in patients and their relatives. These developments will hopefully lead to more effective diagnosis and treatment for affected individuals.⁸

New medications and treatment strategies for rheumatoid arthritis (RA) has led to a decreased need for total joint replacement.¹⁵ However, studies show that patients are frequently non-adherent to prescribed RA medications.¹⁶ As such, family physicians play an integral role toward improving outcomes of RA. As suggested by the CME outcomes data, family physicians need continuing medical education to increase their ability to diagnose RA, and to develop effective treatment strategies. Early use of disease-modifying antirheumatic drugs (DMARDs) markedly reduces inflammation and joint destruction associated with RA. Therefore, with early diagnosis of RA and appropriate use of DMARDs, there is a window of opportunity to change the clinical course of this disabling disease.¹⁷ However, diagnosing RA in the early stages of the disease is difficult.

Studies demonstrate the efficacy of adjusting RA treatment in response to disease activity; however, patients are not always treated according to evidence-based recommendations.¹⁸ Some research also suggests that a *Treat-to-Target Strategy* (T2T) in RA can potentially transform the clinical management of RA into a standardized approach, with the goal of improving both short and long-term goals.¹⁹ Clinical trials have demonstrated that physician-learners would benefit from evidence-based recommendations for implementing T2T best practices, including strategies for overcoming some of the documented T2T implementation barriers.²⁰

Physicians may improve their care of patients with connective tissue disorders by engaging in continuing medical education that provides practical integration of current evidence-based guidelines and recommendations into their standards of care, including, but not limited to the following:^{4,5,21-24}

- Patients with significant internal organ involvement are often asymptomatic until the late stages of systemic sclerosis; therefore, routine monitoring for underlying disease is essential after the initial diagnosis.
- Doppler echocardiography, pulmonary function testing, and high-resolution computed tomography of the chest should be performed at diagnosis of systemic sclerosis and at regular intervals thereafter.
- Treating active interstitial lung disease with oral cyclophosphamide (Cytoxan) for one year modestly improves lung function, dyspnea, skin thickening, and health-related quality of life in patients with systemic sclerosis.



- Initiation and continuation of angiotensin-converting enzyme inhibitors are recommended in patients with scleroderma renal crisis, even in the presence of elevated creatinine levels.
- The gold standard for diagnosing SLE is a rheumatologist's diagnosis. The American College of Rheumatology (ACR) uses a standard classification scheme requiring 4 of 11 criteria for research definition, although this is recognized to miss early and mild cases.
- Treatment for SLE consists primarily of immunosuppressive drugs (e.g., hydroxychloroquine [Plaquenil] and corticosteroids [prednisone]). (1,2) In 2011 the FDA approved the first new drug for lupus in more than 50 years—belimumab [BENLYSTA®].
- The ACR recommends ANA testing in patients who have two or more unexplained signs or symptoms (see guidelines for list of symptoms). Because of the high rate of false positive ANA titers, testing for systemic lupus erythematosus with an ANA titer or other autoantibody test is not indicated in patients with isolated myalgias or arthralgias in the absence of these specific clinical signs. Under most circumstances, a persistently negative ANA titer (less than 1:40) can be assumed to rule out systemic lupus erythematosus.
- A normal-range ANA titer in the context of organ system involvement that suggests systemic lupus erythematosus should prompt a work-up for alternative diagnoses. If no other cause is identified, the diagnosis of ANA-negative systemic lupus erythematosus and consultation with a rheumatologist should be considered. If patients with a normal ANA titer develop new clinical features that are consistent with systemic lupus erythematosus, ANA testing should be repeated.
- Primary muscle weakness must be distinguished from the more common conditions of fatigue and asthenia.
- If the diagnosis is still inconclusive after the history, physical examination, and laboratory, radiologic, and electromyographic evaluation, a muscle biopsy is required for patients who have a suspected myopathy.
- According to a guideline from the College of American Pathologists (CAP), no further laboratory tests are necessary in patients who meet diagnostic criteria for systemic lupus erythematosus and also have a positive ANA test result.
- Testing for antibody to double-stranded DNA antigen (anti-dsDNA) and antibody to Sm nuclear antigen (anti-Sm) may be helpful in patients who have a positive ANA test but do not meet full criteria for the diagnosis of systemic lupus erythematosus. AntidsDNA and anti-Sm, particularly in high titers, have high specificity for systemic lupus erythematosus, although their sensitivity is low. Therefore, a positive result helps to establish the diagnosis of the disease, but a negative result does not rule it out. The CAP guideline recommends against testing for other autoantibodies in ANA-positive patients, because there is little evidence that these tests are of benefit.
- The ACR recommends that primary care physicians consider a rheumatology referral for patients with characteristic signs and symptoms of systemic lupus erythematosus (see guidelines for a list of characteristics), and a positive ANA test, particularly if these patients have more than mild or stable disease.
- Patients with inflammatory joint disease should be referred to a rheumatology subspecialist, especially if symptoms last more than six weeks.
- In persons with RA, combination therapy with two or more disease-modifying antirheumatic drugs is more effective than monotherapy. However, more than one



biologic agent should not be used at one time (e.g., adalimumab [Humira] with abatacept [Orencia]) because of the high risk of adverse effects.

- A guided exercise program can improve quality of life and muscle strength in patients with RA.
- Cardiovascular disease is the main cause of mortality in persons with RA; therefore, risk factors for coronary artery disease should be addressed in these patients.
- Methotrexate monotherapy demonstrated statistically significant and clinically relevant improvement of symptoms and physical function compared with placebo at 12 to 52 weeks.
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Physicians can improve patient satisfaction with the referral process by using readily available strategies and tools such as, improving internal office communication, engaging patients in scheduling, facilitating the appointment, tracking referral results, analyzing data for improvement opportunities, and gathering patient feedback.^{25,26}

These recommendations are provided only as assistance for physicians making clinical decisions regarding the care of their patients. As such, they cannot substitute for the individual judgment brought to each clinical situation by the patient's family physician. As with all clinical reference resources, they reflect the best understanding of the science of medicine at the time of publication, but they should be used with the clear understanding that continued research may result in new knowledge and recommendations. These recommendations are only one element in the complex process of improving the health of America. To be effective, the recommendations must be implemented. As such, physicians require continuing medical education to assist them with making decisions about specific clinical considerations.

Resources: Evidence-Based Practice Recommendations/Guidelines/Performance Measures

- Diagnosis of systemic lupus erythematosus²¹
- Systemic sclerosis/scleroderma: a treatable multisystem disease⁴
- American College of Rheumatology. Clinical Practice Guidelines²⁷
- Evaluation of the patient with muscle weakness²²
- Predicting rheumatoid arthritis risk in adults with undifferentiated arthritis¹⁷
- Diagnosis and management of rheumatoid arthritis²³
- PUTTING EVIDENCE INTO PRACTICE: Methotrexate Therapy for Rheumatoid Arthritis²⁴
- ACR recommendations for the use of disease-modifying antirheumatic drugs and biologic agents in the treatment of rheumatoid arthritis²⁸
- ACR Choosing Wisely²⁹
- Adding health education specialists to your practice³⁰
- Envisioning new roles for medical assistants: strategies from patient-centered medical homes³¹
- The benefits of using care coordinators in primary care: a case study³²
- Engaging Patients in Collaborative Care Plans³³
- The Use of Symptom Diaries in Outpatient Care³⁴



- Health Coaching: Teaching Patients to Fish³⁵
- Medication adherence: we didn't ask and they didn't tell³⁶
- Encouraging patients to change unhealthy behaviors with motivational interviewing³⁷
- Integrating a behavioral health specialist into your practice³⁸
- Simple tools to increase patient satisfaction with the referral process²⁵
- Juvenile Rheumatoid Arthritis | Overview (patient education)³⁹
- Rheumatoid Arthritis | Overview⁴⁰
- FamilyDoctor.org. Scleroderma | Overview (patient education)⁴¹
- FamilyDoctor.org. Raynaud's Disease | Overview (patient education)⁴²

References

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