**Body System:** Musculoskeletal

**Session Topic:** Connective Tissue Disorders

| Educational Format | | Faculty Expertise Required |
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| **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. Please describe your interest and plan for teaching a PBL on your proposal form. |

| Professional Practice Gap | Learning Objective(s) that will close the gap and meet the need | Outcome Being Measured |
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| • Underdiagnosis of Systemic lupus erythematosus (SLE) because the presenting symptoms and signs are often not specific.<br>• Overdiagnosis of SLE because doctors mistakenly use a positive blood test (present in 5% of the healthy population) by itself to make a diagnosis.<br>• Compliance with prescribed treatment is often a problem, given the use of strong immunosuppressive medications and resulting side effects in young, reproductive age women who want to partner and have children. This is a relatively unique demographic/therapeutic problem among the rheumatic diseases.<br>• Patients with lupus have an increased frequency of related autoimmune problems, such as Sjogren's syndrome (i.e., dry eyes, dry | 1. 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.<br>2. 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.<br>3. Counsel patients on treatment regimens to manage symptoms including combinations of immunosuppressant and/or anti-inflammatory medications, diet, lifestyle, and follow-up appointments.<br>4. Establish protocols to recognize and manage possible complications associated with connective tissue disorder treatment. | Learners will submit written commitment to change statements on the session evaluation, indicating how they plan to implement presented practice recommendations. |

 

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| mouth) and antiphospholipid syndrome (i.e., clotting problems, strokes, fetal loss), that require additional treatments. | | |

| **ACGME Core Competencies Addressed** (select all that apply) | | | |
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| X | Medical Knowledge | | Patient Care |
| | Interpersonal and Communication Skills | | Practice-Based Learning and Improvement |
| | Professionalism | | Systems-Based Practice |

**Faculty Instructional Goals**

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.

- 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 References section below are a good place to start
    - 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.

**Needs Assessment:**

*Note – in terms of scope of this education, rheumatoid arthritis is being covered as a separate topics; therefore, this topic should focus more on systemic lupus erythematosus (SLE); scleroderma (Scl); polymyositis (PM); and dermatomyositis (DM).

Dermatomyositis is a rare disease with an incidence of 9.63 per 100,000 (including all subtypes).[1] Systemic sclerosis 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.[2]

 

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.[3]

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.[4] More specifically, CME outcomes data from 2012 AAFP 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.[5] 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]

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:[2,3,6,7]

- 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.

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.[8,9]

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 erythematosus[6]
- Systemic sclerosis/scleroderma: a treatable multisystem disease[2]
- American College of Rheumatology. Clinical Practice Guidelines[10]
- Evaluation of the patient with muscle weakness[7]
- Adding health education specialists to your practice[11]
- Envisioning new roles for medical assistants: strategies from patient-centered medical homes[12]
- The benefits of using care coordinators in primary care: a case study[13]
- Engaging Patients in Collaborative Care Plans[14]
- The Use of Symptom Diaries in Outpatient Care[15]

 

- Health Coaching: Teaching Patients to Fish[16]
- Medication adherence: we didn't ask and they didn't tell[17]
- Encouraging patients to change unhealthy behaviors with motivational interviewing[18]
- Integrating a behavioral health specialist into your practice[19]
- Simple tools to increase patient satisfaction with the referral process[8]
- FamilyDoctor.org. Scleroderma | Overview (patient education)[20]
- FamilyDoctor.org. Raynaud's Disease | Overview (patient education)[21]

## References


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**10.** American College of Rheumatology. Clinical Practice Guidelines. 2014; http://www.rheumatology.org/Practice/Clinical/Guidelines/Clinical_Practice_Guidelines/ . Accessed October, 2014.

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**16.** Ghorob A. Health Coaching: Teaching Patients to Fish. *Family practice management.* 2013;20(3):40-42.

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