



## An uncommon syndrome makes us reflect on our approach to diagnosis

In this issue of the *Journal*, Dr. Soumya Chatterjee and colleagues (page 655) discuss the antisynthetase syndrome. Although uncommon, this syndrome is important for internists and subspecialists to be aware of. Patients present in several different ways, and potentially life-threatening organ involvement may initially not be recognized or may not be linked with other components of the syndrome, such as involvement of the lungs, muscles, heart, and esophagus and fever.

I am currently on our inpatient rheumatology consultation service, and so I am reminded daily of the challenges hospitalists and subspecialists confront in ordering tests while trying to balance limiting length of stay with cost-efficiency and the desire to obtain a correct diagnosis. And I am repeatedly sensitized to several common test-ordering pitfalls intrinsic to the evaluation of patients with multisystem disease, including myositis. Most have a shared theme—limited time is spent in thoughtful reflection before ordering.

Patients with myositis rarely present with the textbook description of proximal muscle weakness. They describe fatigue, malaise, and sometimes a generalized sense of weakness. It is the probing questioning of their functional capacity and focused examination that reveal that the weakness is characterized by difficulty getting up off the floor, out of a low chair, or off the toilet. Then, with further questioning, some patients note that their fatigue and tiredness may also include getting winded easily with exertion, such as when climbing stairs, thus raising the question of cardiac dysfunction, pulmonary hypertension, or interstitial lung disease.

The responses to those probing questions and the subsequent examination should transform the interpretation of elevated aminotransferase levels (“liver tests”: AST and ALT) from liver disease into suspicion of muscle disease and the appropriate ordering of the creatine kinase level (avoiding liver imaging and hepatology consultation). The carefully repeated and now focused neurologic examination distinguishes the initial “poor cooperation” from the proximal weakness of myopathy. The probing interview leads to the performance of a focused physical examination that frames the appropriate interpretation of the routinely obtained “admission lab studies”!

The thoughtful history and examination are the basic stuff of clinical medicine that can easily be pushed aside by any of us as we deal with the tensions of high-volume, “high-throughput” medical care. It is a low-resistance path from hearing the symptom of fatigue with elevated “liver enzymes” to immediately checking ferritin, ceruloplasmin, and a hepatitis screen in preparation for getting a liver biopsy. It is easy to go through the motions without reflection. Easy, but sometimes wrong. And it is just as easy (but likely to be costly and unhelpful) to identify a patient prematurely with “possible autoimmune disease” and to immediately order a panoply of antinuclear and autoimmune serologies, including the Jo-1 autoantibody test.

As Dr. Chatterjee et al point out, we must continuously reflect on our diagnoses, for even after we navigate the pitfalls and avoid missing the diagnosis of myositis, if

we don't continuously assess all the patient's symptoms, repeat the examination in a directed manner, and *then* look for circulating Jo-1 antibody when appropriate, we may well miss the opportunity to recognize that our patient's ongoing fatigue with exertion is a reflection of the well-described association of myositis with interstitial lung disease (which may warrant a change in therapy), and not steroid myopathy or just poor conditioning.

Alternatively, in evaluating a patient who describes a year of feeling tired, suffering generalized muscle pains with low-grade fevers with temperatures of 99.8°F, and total exhaustion for 3 days after cleaning the oven, testing for antinuclear antibodies, extractable nuclear antigen antibodies, and a "vasculitis panel" in anticipation of a rheumatology consultation is not likely to be useful therapeutically or diagnostically.

Despite the daily pressures, we need to keep ourselves grounded in the fundamentals of clinical care: careful listening, purposeful examination, and directed use of laboratory tests and imaging. The downstream consequences of ordering tests for the sake of efficient throughput are quite real, and thoughtful test ordering is one step toward quality care, as well as cost-effective care.

In future months, the *Journal* will delve more deeply into test ordering when, in a joint effort with the American College of Physicians, we will be discussing the use and misuse of specific tests.



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