Diagnosis Labeling: When Treatment is the Problem
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Story from the Front Lines:
A 64-year-old gentleman with a long history of bilateral ptosis, diplopia, and dysphagia in the context of remote facial trauma after a motor vehicle accident presented to his primary physician with complaints of weight gain, painful edema of all four extremities, and a recent compression fracture with resultant back pain. Many years ago, he was diagnosed with myasthenia gravis at an outside facility, though complete records had never been obtained. After initially being treated with immunosuppressive agents, he was lost to follow-up and had continued to work for seven years without medications as an airport baggage tagger. Seven years after his last use of prednisone, he had been seen in follow-up shortly after a 3-day hospitalization for community-acquired pneumonia where he complained of persistent dyspnea on exertion and fatigue. The evaluating physician started him on pyridostigmine and placed a referral to Neurology Clinic.

Over the past several months he had seen a number of different clinicians and his doses of pyridostigmine, prednisone, and azathioprine had all been incrementally increased despite no significant change in his symptoms or objective improvement in his clinical exam. He reported that he didn’t notice much change in his symptoms when he previously took immunosuppressive agents, but overall “felt better” on prednisone.

Nearly 2 years later his medication list had grown from 4 to well over 20. He had seen a variety of specialists and undergone multiple tests to evaluate his persistent symptoms that were refractory to immune-modulating therapies. After 18 months of aggressive medical treatment, he had noticed no significant change in his initial symptoms and now had developed new symptoms that could all be attributed to the adverse effects of corticosteroids: 40 lbs. weight gain, painful bilateral upper and lower extremity edema, compression fracture, proximal weakness, and increased frequency of respiratory infections.

Ultimately disease confirmation was sought and serologic evaluation was negative for both acetylcholine receptor and MuSK antibodies. Electromyogram with repetitive stimulation was negative for any significant decrement or post-exercise increment. Needle examination of the proximal muscles demonstrated non-irritative myopathy.

Teachable Moment:
Overdiagnosis occurs when people without disease are labeled with a disease from which they do not suffer symptoms or from which they will not succumb to an early death. The downsides to overdiagnosis include the negative effects of unnecessary labeling, the harms of unneeded tests and therapies, and the opportunity cost of wasted resources that could be better used to treat or prevent genuine illness.
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Americans assume that more medical care must lead to improved health and well-being, but there is growing evidence that more harm may be the unintended consequence.⁴

The patient presented here unfortunately suffered the consequences of too much medical treatment as the result of well-intended efforts to better the control symptoms of a disease he never actually had. Blinded by the label of a rare disease, his physicians were unable to attribute his fatigue and dyspnea on exertion to a recent admission for pneumonia – the far more probable explanation. Rather than reassure and follow him as he completed his antibiotics, he was started on medications for a diagnosis that existed only in the medical record. The problem grew as well-meaning clinicians continued to increase his medication list without considering the possibility of stopping medications that provided no obvious benefit to the patient.

The rationale for his diagnosis of myasthenia gravis remains elusive. It appears, however, that he has suffered from steroid-induced myopathy and a compression fracture – known side effects of prolonged corticosteroid use, which have led to his inability to continue employment. His bilateral ptosis and diplopia are likely the result of the facial trauma he suffered after a remote motor vehicle accident and the multiple reconstructive surgeries since. While steroids are commonly prescribed for a variety of illnesses, as physicians we can easily forget that their risks can be as dangerous and impactful as their potential benefits. Just as we have made strides in the last several years to guard against polypharmacy—particularly in our elderly patients—now is the time to set our sights on the upstream problem of overdiagnosis.

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References: