

Transcript Details

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MS Misdiagnosis: Understanding the Frequency and Consequences

Announcer:

Welcome to *NeuroFrontiers* on ReachMD. On this episode, we'll discuss the misdiagnosis of multiple sclerosis with Dr. Andrew Solomon. Not only is Dr. Solomon a neurologist at the University of Vermont Medical Center and an Associate Professor at the Larner College of Medicine at UVM in Burlington, Vermont, but he also presented a session on this exact topic at the 2024 Congress of the European Committee for Treatment and Research in Multiple Sclerosis. Here's Dr. Solomon now.

Dr. Solomon:

This session was part of a larger session focused on the differential diagnosis of MS or disorders that are frequently mistaken for MS, and my particular talk was focused on the frequency of misdiagnosis of multiple sclerosis, so disorders that are in the differential diagnosis for MS and are actually mistaken for MS.

It's important to clarify what we mean by misdiagnosis of MS when we talk about this. What we don't mean is a patient where somebody said, "Well, I think you might have MS" or "I'm worried about MS," and they're sent to a neurologist for further evaluation, and they are found not to have MS. That wouldn't really be considered a misdiagnosis per se, especially if the initial provider who raised concern wasn't even a neurologist, which is often the case. Instead, what we're talking about are patients who are given a diagnosis of MS. They're told they have a diagnosis of MS. They're frequently on treatment for MS with disease-modifying therapy, and they are subsequently evaluated, and it is determined that they in fact do not have MS. They have another diagnosis.

In this presentation, I discussed 10 studies that have been performed since 2019 that have looked at misdiagnosis in subspecialty clinics. They've looked at the number of patients that they've seen who came with an existing diagnosis of MS where that diagnosis was reversed.

These data are in diverse regions. They include subspecialty MS centers in the United States, Portugal, Spain, Iraq, Peru, Argentina, Netherlands, Sweden, and Brazil, so we have a sense that misdiagnosis of MS is quite frequent from these studies.

What these studies have done is they've looked at over a time period the number of patients who were referred to their center with an existing diagnosis of MS, not a question about the diagnosis of MS, and they looked at the proportion of those patients who had this existing or ongoing diagnosis of MS that was reversed and had been misdiagnosed. And the percentage of patients that they found ranges from 5 percent to even 46 percent at one site over varying timeframes. Some of these centers looked over just a year of referrals. Some looked over a much longer period of time, like eight years.

Although these studies all really performed in the last six years or so suggest that misdiagnosis of MS is a very frequent problem in our field, these sort of studies have important limitations. And perhaps most importantly is referral bias, right? So these are patients who are referred to a subspecialty center, so they don't really give us a sense of what's going on out in the community in areas where there aren't subspecialists. In many cases, these are misdiagnoses of MS made by a different provider than the provider who uncovered the misdiagnosis and reversed the diagnosis of MS, so that limits how well we can really assess what caused it because we don't have the records often or the provider there to understand what their thinking was that led to a misdiagnosis.

So for instance, the disorders that were found mistaken for MS most frequently were common disorders like migraine or functional neurologic disorder with abnormal MRI findings like small vessel ischemic disease, and these are disorders that are frequently referred for MS evaluation to begin with, so it's likely that sometimes they will get misdiagnosed; but on the other hand, there are disorders like NMO spectrum disorder which are less prevalent but maybe proportionally mistaken for MS much more frequently given their clinical overlap, so we're not really sure from this data what disorders are the most frequently mistaken for MS.

Misdiagnosis of MS has important consequences for patients, and certainly, this should be obvious. It's not good for anybody to be misdiagnosed. But in some of these studies, patients who have been incorrectly given a diagnosis of MS carried that diagnosis for a long time. In one study, a third of these patients carried that diagnosis longer than 10 years, and the majority of these patients are on MS disease-modifying therapies, which are associated with known risks, including a potentially fatal risk of PML. In one study, 31 percent of patients who had been misdiagnosed with MS suffered unnecessary morbidity as a direct result of misdiagnosis, according to their assessing neurologists. And in several studies, we see patients who don't have MS who participated in MS clinical trials for new therapies, which is an exceptionally high unnecessary risk for them. Coupled with this we also know that these patients are not being treated for their correct disorders, so they may continue to suffer from symptoms of another disorder that has not been correctly diagnosed, so there's important consequences for patients who are misdiagnosed with MS.

Announcer:

That was Dr. Andrew Solomon discussing his presentation at the 2024 Congress of the European Committee for Treatment and Research in Multiple Sclerosis, which focused on the misdiagnosis of multiple sclerosis. To access this and other episodes in our series, visit *NeuroFrontiers* on ReachMD.com, where you can Be Part of the Knowledge. Thanks for listening!