

BroadcastMed | mac_010441_autoimmune_disease_associated_with_testicular_cancer-1080p.mp4

SEAN PITTOCK: We have identified a new form of paraneoplastic encephalitis. This disorder causes ataxia or imbalance, dizziness, double vision, hearing loss essentially due to inflammation of the back of the brain, called the brain stem or the cerebellum. These patients are all men, because the biomarker, the Kelch 11 antibody, is a biomarker of testicular cancer.

So in the neuroimmunology laboratory, we've made many seminal discoveries over the years. And they all stem from the staining pattern of patient antibody on mouse brain slices. And every week, we see about 10 to 15 new patterns of staining. And all those patterns mean something very important.

Now, sometimes what happens is we start to identify the same pattern over and over again. And when we ring the doctors and we start to get the same story over and over again, then we know that we have discovered a biomarker. This is how we discovered this sparkles pattern, like stars shining in the night.

We rang the doctor. And we were told that this was a man with ataxia who turned out to have a testicular cancer. And then, a year later, we saw another one. We call the doctor. Same story.

DIVYANSHU One of the challenges to diagnose these syndromes or diseases is early diagnosis of these conditions because of
DUBEY: the variable presentations they come up with. We are starting to learn more and more about these diseases with the biomarkers, such as Kelch 11, which can help us do a serological test to diagnose them.

But early on, when they present, it's hard to put them in a basket. The other problem which sometimes we have is our ancillary test, such as brain MRIs or even sometimes CSFs, don't give us a clear obvious answer, that this is a paraneoplastic disease.

SEAN PITTOCK: In autoimmune neurological disorders, in my experience, most patients that don't get to centers of excellence don't get a diagnosis.

DIVYANSHU If the disease goes undiagnosed, the inflammation continues to brew. And we have come across some patients
DUBEY: who were diagnosed years and years after the early initial onset of the inflammation inside the brain. And given the debilitating nature of the disease, if not diagnosed early, it reaches a level of irreversibility.

SEAN PITTOCK: At Mayo Clinic, we've recently developed an autoimmune movement disorder evaluation. So, for example, if a patient comes into the doctor's clinic and they have a movement disorder, the doctor can think, well, this could be immune-mediated. It could also be genetically. It could also be related to a metabolic abnormality.

But one of the differential diagnoses is an immune-mediated disorder. And therefore, the physician can order an autoimmune encephalopathy or an autoimmune movement disorder evaluation. And contained in that evaluation are antibodies that are pertinent to that condition.

And so Kelch 11 antibody will be an antibody that will be added into that evaluation. And at the current time, Dr. Dubey, myself, and our team are actually developing a test that can be utilized easily and quickly to assist in making that diagnosis.

DIVYANSHU This will allow us to diagnose these patients early when they present. This will help us, following the diagnosis, to
DUBEY: treat these patients early to bring about the maximum amount of recovery possible. It will also allow us to learn and study this disease further to figure out more individualized treatment so that we can help these patients and reduce the amount of risk associated with these immunotherapies.

We are likely to find more and more of these patients because of the discovery of this particular antibody or biomarker. In a population-based study, we've already estimated that there are about 4,000 to 4,500 patients in the United States who probably have the syndrome.

SEAN PITTOCK: The potential for the ability to look for antibodies in patients' serum that target the entire human protein will have huge impact towards understanding the immunological signatures of cancers. And I think that this is really the start of something great. And I think that this is a great time for patients with autoimmune neurological disease.