

Transcript Details

This is a transcript of an educational program. Details about the program and additional media formats for the program are accessible by visiting: <https://reachmd.com/programs/neurofrontiers/uncover-tk2d-genetic-testing-diagnostic-challenges/70167/>

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www.reachmd.com
info@reachmd.com
(866) 423-7849

Uncovering TK2d: How to Navigate Diagnostic Challenges

Announcer:

You're listening to *NeuroFrontiers* on ReachMD. On this episode, we'll hear from Dr. Kaitlin Batley, Director of the Pediatric Neuromuscular Disease Program at Children's Health and an Assistant Professor of Pediatrics and Neurology at UT Southwestern. She'll be discussing how genetic testing can help us diagnose thymidine kinase 2 deficiency, or TK2d. Here's Dr. Batley now.

Dr. Batley:

We're so fortunate to work in an era where genetic testing is readily available for many patients in our country. And so I think the important thing is to really know what's on the panels that you're ordering and what's covered by the testing that you've chosen. I think we've all been at certain points where we thought a gene was on a panel and then realized later it wasn't, so knowing exactly what the panel that you've chosen includes or whether this is a patient where you would jump to something like a whole exome sequence.

And so because TK2d is autosomal recessive inheritance, we know that we're looking for pathogenic variants in TK2, and sometimes we can get genetic testing that might be inconclusive. Maybe we have one pathogenic variant and one variant of uncertain significance, and that can require a little bit more investigation. And so in those instances, that's where we really look to our slightly more invasive but still really informative tests such as electromyography and nerve conduction studies, as well as muscle biopsy.

And so when we are able to confirm the diagnosis using our other ancillary tests, then that can help us get the correct diagnosis for the patient so that ultimately, we're able to pursue appropriate treatment and can also help us to understand this condition more fully. I do think that this is likely to be an underdiagnosed condition, particularly for those individuals who have the later onset, more slowly progressive form of the disease. And so this can help us understand them a little bit better as well.

So considering the muscle biopsy when appropriate can be really important, and also remember that things like creatine kinase level, which is a readily available blood test, can also help point us in the right direction. And characteristic MRI findings can sometimes be helpful as well. And so we do have really a large number of tests that we can perform to help us get at the correct diagnosis, and I think that working through those is crucial.

Sometimes, we do have to utilize kind of everything that we have in our arsenal in order to make the correct diagnosis. The differential diagnosis when you're working up a patient who has symptoms of TK2d is very broad. This condition can really mimic numerous other neuromuscular mitochondrial diseases. And so I think, fortunately, in the age of genetic testing, we are able to test for many of these things at once. Genetic testing alone can be just crucial.

Again, this comes down to knowing what's on the particular panel that you've ordered and knowing exactly what you've already tested for and what might be missed by those tests. For instance, facioscapulohumeral dystrophy is not a disorder that would be picked up on your panel testing. And so this has to be assessed for separately, and there can be a lot of common symptoms with striking facial weakness and some proximal muscle weakness as well. And so knowing that different tests have different purposes, when to expand our testing, and when to move on beyond genetic testing to really rule out some of these differential diagnoses, some of which, like spinal muscular atrophy, have their own disease-modifying therapies, I think the most important thing is to really pursue getting that final diagnosis for each patient and to pursue testing that might span different modalities if needed.

Announcer:

That was Dr. Kaitlin Batley discussing how genetic testing helps inform TK2d diagnosis. 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!