

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/recognize-signs-tk2-deficiency/70166/>

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Recognizing the Signs of TK2 Deficiency

Announcer:

Welcome 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 the importance of recognizing and diagnosing thymidine kinase 2 deficiency, or TK2d. Here's Dr. Batley now.

Dr. Batley:

Because TK2d can be so highly variable and many of the symptoms can be nonspecific, I think it's important to keep TK2d on the differential in general whenever you're working up a patient who has any type of neuromuscular weakness. That can include the axial and appendicular, and that can be facial weakness with ptosis and ophthalmoplegia. I think any of those signs, if present, should prompt you to make sure that TK2d has been evaluated for.

It's somewhat nonspecific, but it should really be considered because now there is an approved therapy. And so I think whenever we have a diagnosis that has a particular intervention, that's something that's extra important for us to include, but also to begin these patients on the appropriate supportive management as well, which we know is beneficial if we begin earlier in the disease course.

And so with that, the diagnosis should really be considered for any patient who's presenting with neuromuscular or mitochondrial-type symptoms. This is an ultra-rare condition but is likely under-recognized, and I do think that as our awareness of this condition increases, we are likely to find this diagnosis more commonly than we thought. These kinds of recognition are so important because when we do have an available FDA-approved treatment for a condition, it has to be added to our do-not-miss list. Getting the diagnosis is always crucial, but when you have a therapy that could potentially modify disease, it becomes even more so. And so knowing that this is one that we need to rule out or assess for is really important.

Announcer:

You just heard Dr. Kaitlin Batley discussing the importance of recognizing and diagnosing TK2d. 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!