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## Recognizing TK2d Across the Lifespan: Clinical Clues by Age of Onset

### Announcer:

This is *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 the clinical presentation and progression of thymidine kinase 2 deficiency, or TK2d, vary based on age of onset. Here's Dr. Batley now.

### Dr. Batley:

The clinical phenotype of TK2d can really vary depending on the age of onset, and I think that's one of the things that makes this condition somewhat tricky to diagnose. It really can have some nonspecific symptoms that can mimic many other much more common disorders.

So, for instance, in the infantile onset form of the disease, there is often a rapidly progressive respiratory impairment, significant hypotonia, and muscle weakness, and sometimes this can look similar to spinal muscular atrophy or different congenital myopathies or muscular dystrophies. So that can confuse the diagnosis.

Then, in somewhat older children, after infancy and before adolescence, we have a more childhood-onset form of the disease whereby common symptoms can include hypotonia, gross motor delay, regression, or loss of motor milestones, or simply with fatigue, exercise intolerance, and proximal muscle weakness.

And so one of the things that can be a little bit more specific for TK2d as far as symptomatology is the striking facial weakness, often with ptosis and ophthalmoplegia as well. And so if you see those symptoms in combination with more systemic proximal predominant muscle weakness, that can really point you towards TK2d.

And then I think probably the most challenging group to diagnose and probably the most underdiagnosed in general would be the later-onset phenotype. And so this can be later adolescents and teenagers all the way up through adulthood, and these individuals can have more subtle, slowly progressive muscle weakness that's predominantly proximal, as well as the fatigue and some of the bulbar and ophthalmoplegia and ptosis signs as well.

Age of onset in TK2d in particular is really crucial to understanding the phenotype and to really get you to the correct diagnosis. And so understanding that the time period in which a patient presents with symptoms will really dictate how severe their symptoms are and how rapidly they progress. We know that those patients with a more severe phenotype will present in infancy shortly after birth and will have the most severe symptoms, often rapidly progressive and leading to mortality typically within one year of life. And then those who have a more subtle or more mild phenotype may not present until later on in adulthood, and their symptoms are much more slowly progressive and can include proximal muscle weakness and fatigue.

### Announcer:

That was Dr. Kaitlin Batley discussing the variable presentation and progression of 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!