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/optimizing-tk2d-care-through-a-multidisciplinary-approach/29877/

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

Optimizing TK2d Care Through a Multidisciplinary Approach

Announcer:
You're listening to *NeuroFrontiers* on ReachMD, and this episode is supported by UCB. Here's your host, Dr. Charles Turck.

Dr. Turck:
This is *NeuroFrontiers* on ReachMD, and I'm Dr. Charles Turck. Joining me to share their insights on multidisciplinary care for thymidine kinase 2 deficiency, or TK2d for short, are Drs. Kaitlin Batley and Esra Caylan. Dr. Batley is the Director of Pediatric Neuromuscular Medicine at Children's Health and an Assistant Professor at UT Southwestern Medical Center. Dr. Batley, welcome to the program.

Dr. Batley:
Thank you so much. I'm happy to be here.

Dr. Turck:
Also coming to us from Children's Health and UT Southwestern Medical Center is Dr. Caylan, who's a pediatric pulmonologist and an Assistant Professor in the Department of Pediatrics. Dr. Caylan, thanks for being here today.

Dr. Caylan:
Thank you so much for the opportunity and the invitation.

Dr. Turck:
So starting with some background, Dr. Caylan, what are the most common respiratory symptoms you see in patients with TK2d?

Dr. Caylan:
TK2d is a disease that affects muscle strength. When we have a disease affecting muscle strength, it affects our breathing. Our breathing is muscle-dependent because we have our diaphragm as the biggest muscle in our body and then multiple smaller muscles helping us breathe. When muscle strength decreases, our breath becomes less efficient, and we need support. That also affects how strongly we can cough and clear our lungs. When you're not able to clear your lungs efficiently, that will increase your risk of respiratory infections and pneumonia and increase lung damage.

Dr. Turck:
And how about neurological and ocular symptoms, Dr. Batley, which ones do you often see?

Dr. Batley:
So TK2d is a very heterogeneous condition, and the symptoms really can vary depending on the severity in the form as well as age of onset of symptoms. So we know that our infantile-onset patients who tend to develop symptoms before the age of two years old are those who tend to have the most severe manifestations of this disorder. And so in those patients, they may have an initial period with a normal motor exam and development, followed by the onset of fairly rapidly progressive weakness, hypotonia, and loss of these motor milestones.

And so in those patients who are the most severely affected, we can see weakness of the eye muscles leading to abnormal extraocular movements, as well as facial and bulbar weakness. This can then lead to troubles with speaking or dysarthria, as well as difficulties swallowing, leading to dysphasia and feeding difficulties. And in those cases, they can often require things like a feeding tube for supplemental nutrition, as well as assistive devices for communication.

This weakness can also extend to the respiratory muscles, which can lead to neuromuscular respiratory insufficiency and sometimes even respiratory failure, requiring assisted ventilation. And these patients will often have trouble with muscle strength and will require

adaptive devices like wheelchairs in order to optimize mobility.

In patients who have later-onset symptoms—like those with juvenile or adult onset—the symptoms can be milder and a bit more nonspecific, with progressive motor weakness that tends to be more proximal. And in these symptoms, there can be kind of a variety of things that people notice. Adults, for instance, might notice more difficulties with things like getting up from a chair or climbing up stairs, but it can be subtle and therefore more difficult to diagnose. But the findings of extraocular movements, abnormalities, or eye muscle weakness are also key features of TK2d, especially in those with later-onset disease.

Dr. Turck:
Now, staying with you for just another moment, Dr. Batley, would you tell us about some diagnostic challenges you've experienced with TK2d and how a multidisciplinary approach might help mitigate them?

Dr. Batley:
Because of the variability of symptoms, this can be a difficult disease to diagnose, and we do suspect that overall it is underdiagnosed, especially in those individuals who have milder and later-onset symptoms. And so this is when the multidisciplinary team is incredibly important in order to make sure that we're identifying these patients who do happen to have more subtle symptoms. So for instance, a patient might experience more significant respiratory problems than motor function problems. And so they might first present to a pulmonologist. And in that way, the pulmonologist is able to use their expertise to say, “This looks like it could be a neuromuscular respiratory insufficiency; you really need to see a neurologist or a geneticist,” and then they might be able to get them to the appropriate care. And, for instance, other features of the disease, such as hearing difficulties, might lead them to see an audiologist first. And similarly, the audiologist might be the one to identify, “Hey, I think this is more of a multisystemic condition,” and get them to the right place.

And so having this multidisciplinary team where we're able to work together and refer to the right person to continue work-up is really crucial in identification, especially for those with more subtle symptoms.

Dr. Turck:
For those just tuning in, you're listening to *NeuroFrontiers* on ReachMD. I'm Dr. Charles Turck and I'm speaking with Drs. Kaitlin Batley and Esra Caylan about how to establish a multidisciplinary approach to thymidine kinase 2 deficiency, or TK2d.

So now that we've discussed the symptoms and diagnosis of TK2d, let's focus on its management. Dr. Caylan, what role do you play on the multidisciplinary care team?

Dr. Caylan:
As pulmonologists, we need to get involved with TK2d patients early, because they will have symptoms early in life or develop them later in life. We need to screen them, give them treatment, and adjust our treatment plans. They usually need invasive or noninvasive ventilation support with a BiPAP, CPAP, or sometimes even a tracheostomy with ventilation. They need airway clearance equipment. They will need some medications. They might need a cough assist to help them with an efficient and strong cough to be able to keep their lungs clear and decrease the risk of respiratory infections and pneumonia.

Dr. Turck:
And Dr. Batley, who else is on the multidisciplinary team? And what are their roles?

Dr. Batley:
So the really unique thing about the multidisciplinary team is that it is very individualized for each particular patient depending on what their particular needs are. And so the team can really vary. I would say a neurologist who has comfort in neuromuscular medicine is a really important part of the care team, especially in my opinion, as one myself. And this is someone who can guide, direct, and identify which other team members might need to be brought in. So of course, a pulmonologist with comfort in neuromuscular physiology is also very crucial. And then other patients may require things like an audiologist to address hearing loss. Sometimes a gastroenterologist, if there are significant feeding difficulties from dysphagia or weakness when swallowing. Also a very important part of the care team are our therapists, so physical, occupational, and speech therapists who can help to both optimize developmental outcomes in younger children as well as assist with things like adaptive technology and assistive devices to make sure that we're supporting each individual patient with what they need in order to really optimize their quality of life. And then things like speech therapy can work both on feeding as well as addressing some of the weakness of the muscles of speaking—which can lead to dysarthria—to help with communication as well.

And so it really is a very individualized team, but it definitely takes many players in order to make sure that we're addressing each individual need for a disorder that affects so many different systems of the body.

Dr. Turck:
Now, before we close, I'd like to ask each of you to share one key recommendation for collaborating with a multidisciplinary team. Dr. Caylan, I'll start with you.

Dr. Caylan:
We are dealing with a very rare disease, so everyone's experience is low because this is not a disease we see every day. So we actually need each other's experience and knowledge about this disease to be able to better understand it and to be able to manage and help our patients better. We need to work as a multidisciplinary team to come together and give a complete approach to help patients.

Dr. Turck:
And Dr. Batley, I'll give you the final word.

Dr. Batley:
Being a part of a multidisciplinary care team is such an amazing opportunity. We're really able to rely on our colleagues who have diverse and unique experiences and expertise. And that's so important when we're working in rare disease where we might all have just a few patients who meet these particular characteristics, and so we're able to really rely on each other to make sure that we're addressing each individual's unique needs.

I think that one of the most important things that we can always keep in mind when we're working on this kind of team is to keep our lines of communication open. Because of the variability of these disorders, it's important that we're able to reach out to each other and address the unique values and goals of care that each patient has. And so good communication and remembering the kind of unique nature of each of our patients are some of the most important things that we can do on our multidisciplinary care teams.

Dr. Turck:
Thank you both for sharing those recommendations. And as that brings us to the end of today's program, I want to thank my guests, Drs. Kaitlin Batley and Esra Caylan, for joining me to discuss multidisciplinary approaches to the diagnosis and management of thymidine kinase 2 deficiency. Dr. Batley, Dr. Caylan, it was wonderful speaking to you both.

Dr. Batley:
Thank you so much. It was great to speak with you as well.

Dr. Caylan:
Thank you so much.

Announcer:
This episode of *NeuroFrontiers* was supported by UCB. 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!