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Optimizing Screening Pathways for Alpha-1 Antitrypsin Deficiency

Announcer:
You're listening to *Clinician's Roundtable* on ReachMD, and this episode is sponsored by Grifols. Here's your host, Dr. Alexandria May.

Dr. May:
Welcome to *Clinician's Roundtable* on ReachMD. I'm Dr. Alexandria May, and joining me to discuss the latest guideline recommendations and diagnostic strategies for alpha-1 antitrypsin deficiency is Dr. Michael Czarnecki. He's a pulmonologist at "The Lung Docs" Pulmonary and Critical Care Consultants in Chattanooga, Tennessee. Dr. Czarnecki, it's great to have you with us today.

Dr. Czarnecki:
Thank you. Looking forward to our conversation.

Dr. May:
Well, let's dive right in, Dr. Czarnecki. Despite longstanding recommendations and accessible testing, alpha-1 antitrypsin deficiency is still underdiagnosed. Based on your experience, why do you think this gap continues to persist in everyday clinical practice?

Dr. Czarnecki:
Well, there certainly is a knowledge gap of awareness, and one of our jobs as pulmonologists and certainly as the community in lung diseases—some people will say a rare lung disease, but what we often recognize is it's a common disease, just rarely tested for—we want to encourage and educate providers, staff, and patients in the community that screening is very important. We want to screen for diseases that may not be commonly tested for and have a low bar for thinking about alpha-1 antitrypsin deficiency. And increased awareness will allow for increased testing and therefore increased diagnosis, increased treatment, and hopefully prevention of worsening disease.

Dr. May:
As a follow-up to that, what are some of the most common assumptions or misconceptions that can prevent clinicians from testing a patient?

Dr. Czarnecki:
Well certainly, one of the most common things I hear is 'you cannot diagnose a genetic condition by looks alone.' It's genetic, so you have to implement some sort of screening protocol or strategy to identify genetic conditions like alpha-1 antitrypsin deficiency into your practice.

The other misconception or myth is not everything that wheezes is asthma. Many patients can proceed many years—on average, up to eight years—before they're diagnosed with alpha-1 antitrypsin deficiency because they've been misdiagnosed with maybe asthma or some other condition. And the reality is you were never screened. So a screening test is very important. You oftentimes can go through up to two providers before you actually get a diagnosis of alpha-1 antitrypsin deficiency. So those are two main misconceptions.

The other that I hear often is, well, we don't have any good treatments for alpha-1 antitrypsin. So you want to not just have a strategy to protocolize testing and screening, but you also want to recognize that there are options. There are very good treatments for alpha-1 antitrypsin deficiency like replacement therapies that augment your immune system to replace the deficiency you have.

Dr. May:
Now, family history can be a powerful but underused tool. So can you tell us about its role when you're identifying patients who may warrant testing?

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Dr. Czarnecki:
When I go out and educate providers and talk to community support groups, there's a very beautiful graphic slide we show of a family tree. And my old infectious disease colleague used to say to me, "When you aren't sure of a disease or what's going on, go back and get more history." And so you want to go get family history, clinical history, and the history of present illness.

So your generation above you—your mom and dad—what kind of breathing problems did they have? Did they die of a condition at an early age? Did they have complications early in life from breathing or heart issues? Were they told that they had a liver disease throughout their life that they struggled with but were never actually screened for some sort of genetic condition like alpha? Did they have any offsprings that died early in neonatal years?

And so these are all pieces of the puzzle you start to put together. Come up with a diagnostic strategy to figure out what's wrong or what condition your patient may or may not have, and allow yourself to think about, 'oh, I should screen this patient for alpha-1 antitrypsin deficiency with a screening test.'

Dr. May:
For those just joining us, this is *Clinician's Roundtable* on ReachMD. I'm Dr. Alexandria May, and I'm speaking with Dr. Michael Czarnecki about how we can better recognize and diagnose alpha-1 antitrypsin deficiency early.

Let's shift gears a bit and focus on the latest screening recommendations. Both the American Thoracic Society and European Respiratory Society Joint Statement *and* the 2025 Canadian Thoracic Society guideline state that all patients with COPD should be tested for alpha-1 antitrypsin deficiency at least once, regardless of age, smoking history, or disease duration. They also highlighted additional groups such as those with liver disease, bronchiectasis, or a family history. With all that being said, Dr. Czarnecki, what does the application of those recommendations look like in your practice?

Dr. Czarnecki:
So when you look at the ATS and ERS criteria or the Canadian guidelines, they're always evolving, and they get updated on a fairly consistent basis. And regarding the latest update, with a broad stroke, I'll sum it up for you: test everyone and screen everyone. Any asthma patient who has not completely reversible airway obstruction in their PFT should be tested. Regarding the other criteria you pointed out, any COPD patient who's never been tested should be tested. Family history of early lung diseases should be tested.

Now, as a primary care physician, you may be faced with some time constraints as well, and you may want to be a little more selective. And that's where you will really take these guidelines now and have to implement some sort of protocol into your practice or workflow. The screening test itself is fairly straightforward. It's a cheek swab for DNA testing that takes less than three minutes to do, and you send it off.

So how do you build a three-minute test into your protocol? And the way we do it—and the way I advocate going around talking to different groups and practices—is one, if you're in a pulmonary group, consider implementing it into your PFT or your spirometry testing. Have your technician do it. Any patient that comes in and gets a PFT is going to get swabbed automatically. That way, you don't miss any patients.

Based upon my experience, we optimized that workflow a little bit better so that we don't miss anyone. And one of the ways we did it was for any patient that is new to our practice, they automatically get screened. Any patient that is leaving my exam room after myself or one of our providers has seen the patient, and we either change their inhaler or we add a new inhaler or some sort of respiratory medicine, then on the way out and during their checkout, they get screened. So that process essentially covers not only everyone, but it satisfies every single point on the guideline criteria that have been updated.

Dr. May:
And once you've applied these guidelines and identified an appropriate patient for testing, how do you go about diagnosing them?

Dr. Czarnecki:
The first step is a screening process with that cheek swab. That gets sent off, and you usually get the results back into your electronic medical record system, faxed to you, or whatever method you use within 10 days. And then, we have a follow-up. For all our patients that have been screened or have a test initiated, we'll have a two- to four-week follow-up, and we then review the results with the patient and determine whether they're a normal patient and they have no genetic deficiency, they're an alpha carrier, or they have severe deficiency.

And then the next step is another follow-up to talk about—after additional testing and a confirmatory test to confirm the screening test—what are the next steps? Do we watch and wait, or do we offer therapy? We educate, and that goes down a whole other pathway of follow-up and consistent follow-up with your provider to ensure there are no declines in your lung function or your clinical health.

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Dr. May:
Before we close Dr. Czarnecki, let's bring all this together. Given the importance of screening and these guideline recommendations, what strategies can help us embed this testing into routine clinical workflows?

Dr. Czarnecki:
One of the most important things that we've found is if you really want to implement this into your practice, you find a champion. So we advocate taking the test kits and screening not just your providers and myself, for example, but all my staff. And you'll be surprised how many people are identified as carriers, and that person becomes a champion for the practice to not just advocate for the patients coming to your practice, but to help implement the protocols that you want to put in place.

But furthermore, let's say no one in your practice is identified as a carrier or severely deficient. Just the mere fact that you're educating your staff opens up the door for a conversation and say, 'Hey, you know what? My uncle or my dad or a family member of mine had this condition,' and boom, there's your champion right there. You can then empower them to be part of not just your practice, but part of patient care. So they become not just an administrator to help patients get in, checked in, checked out, ordered labs, follow-up appointments, and things like that, but now they actually become part of the process of caring for patients and they're empowered to be the champion for your practice.

Dr. May:
Well, those are certainly great strategies for us to bring back to our clinical practices. But as we wrap up today's program, I want to thank my guest, Dr. Michael Czarnecki, for joining me to share these best practices for diagnosing alpha-1 antitrypsin deficiency. Dr. Czarnecki, it was great speaking with you today.

Dr. Czarnecki:
Thank you very much.

Announcer:
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