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

Collaborative Strategies Toward Early Detection of Alpha-1 in COPD

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

Dr. Turck:
Welcome to *Clinician's Roundtable* on ReachMD. I'm Dr. Charles Turck, and joining me to share their perspectives and how we can better collaborate to detect and treat alpha-1 antitrypsin deficiency in patients with chronic obstructive pulmonary disease, or COPD for short, are Dr. Brian Smith and Dr. Charlie Strange. Dr. Smith is in private practice at Versailles Family Medicine in Kentucky. Dr. Smith, thanks for being here today.

Dr. Smith:
Happy to join you.

Dr. Turck:
And Dr. Strange is a Professor of Pulmonary, Critical Care, Allergy, and Sleep Medicine at the Medical University of South Carolina. Dr. Strange, it's great to have you with us.

Dr. Strange:
Nice to be here, Charles.

Dr. Turck:
Starting with you, Dr. Strange, can you tell us how alpha-1 antitrypsin screening fits into COPD care algorithms?

Dr. Strange:
Alpha-1, for short, is the most common genetic cause of COPD. And I think most people don't realize that we have a naturally occurring chemical in our bloodstream called alpha-1 antitrypsin; it's the second most abundant protein in our bloodstream. And alpha-1 antitrypsin is a very anti-inflammatory molecule to a lot of different organ systems, including the lungs. So you think of it as the natural protection of the lung. And when you're genetically deficient in this protein, then other environmental insults to the lungs then cause havoc inside the lower airways. And so that could be cigarette smoke or environmental particulates or fumes. And the combination of having a low alpha-1 level and the insult then produce COPD. This is about 1% of all patients with COPD. And therefore, it's one of our 7,000 rare diseases that's worthy of detection, even at the primary care level.

Dr. Turck:
With that background in mind, let's turn to you now, Dr. Smith. As a primary care physician, how do you implement alpha-1 screening into your practice?

Dr. Smith:
In primary care, when I talk to primary care physicians about screening for alpha-1, a lot of the problems is primary care physicians don't have spirometry in their office. And even if they do, doing a spirometry and then a treatment and then another spirometry test to prove what type of COPD versus asthma that a patient has is time-consuming. And in a primary care practice, it's very difficult to implement. So what I encourage people to do in primary care, especially given that the test is available for free in a variety of ways, just whenever you're prescribing an inhaler, go ahead and order the alpha-1 test on that patient. There's no real harm in doing it. A lot of times if you're drawing blood anyway, you can do it from the blood sample. So it's not an extra stick or anything. And then we also now have, some cheek swabs since it is a DNA test. We can do the genetic tests based on a cheek swab.

Dr. Turck:
And then once you screen patients for alpha-1, Dr. Smith, how and when do you refer them to a pulmonologist?

Dr. Smith:
So there's probably a few instances. We would screen a patient, and then the primary care physician gets the result back and it's abnormal, there's several different abnormalities that you can have with alpha-1, some that can cause disease and some that don't cause disease. And if the primary care physician doesn't want to get lost in the weeds there, any abnormal result I think is reasonable to send to a pulmonologist.

Then also we can take those abnormal results and reach out to that patient's family, since we're in primary care and family medicine, and encourage the rest of the family to get tested. That's how we end up finding a vast majority of our patients is we get one positive, and then that one positive patient spreads through the family tree. And so then we can backtrack and find if there's any homozygous patients to refer on to pulmonology too. Or like I said it can be a little bit confusing, the results. A lot of times if it's just any abnormal result, we would send that on to the pulmonologist.

Dr. Turck:
For those just joining us, this is *Clinician's Roundtable* on ReachMD. I'm Dr. Charles Turck, and I'm speaking with doctors Brian Smith and Charlie Strange about how primary care physicians and pulmonologists can better collaborate to detect and treat alpha-1 antitrypsin deficiency in patients with COPD.

So Dr. Strange, once primary care physicians like Dr. Smith refer patients to you, what are some common challenges you face when coordinating a patient's care?

Dr. Strange:
I think it's important for the audience to recognize that this diagnosis makes up about 1% of all patients with COPD. And the severe deficiency occurs when you have two abnormal alleles on the alpha-1 gene that are involved with deficiency genes. And what that means is that there are another 10% of patients with COPD in most of our studies that are carriers of one abnormal allele. What that means is that those individuals do have extra risks for COPD. So now primary care physicians can discuss with this individual and their family members why they're uniquely susceptible to the environmental insults to the lungs. And it helps with the smoking cessation discussion and all the other pieces of this. So it truly is a collaborative effort to try and recognize severe deficiency and then the carrier state because both of those have clinical implications for families. Getting all of that information in the hands of the patient and in the primary care referral practice is really, really important.

Dr. Turck:
Now we're almost out of time for today. So before we close, are there any best practices or strategies either of you use to enhance a patient's care coordination? Dr. Smith, I'll start with you.

Dr. Smith:
I think it's important to find pulmonologists in your area that will treat alpha-1 deficiency, and then work closely with them. And then also having that feedback from the pulmonologist; there is something that they're doing different for that patient. It's very helpful for us to have some information, feedback, and encouragement to continue testing for a rare disease. Because sometimes that can be a little frustrating. I mean, you get all these normal results in a row, which is good. But then you're like, 'Well, is this really out there?' And so having that feedback with the abnormal results and that there's something that can be done for them is very helpful.

Dr. Turck:
Thanks, Dr. Smith. And Dr. Strange, I'll give you the final word.

Dr. Strange:
I think this is an important diagnosis for primary care. The challenge here is the repeated testing to find the individuals at risk. But once you identify one person, our job as a specialist is to return that patient back to primary care because the therapies that are employed for alpha-1, although there are many in the pipeline right now, are weekly intravenous infusions that can be monitored and supervised by the primary care team as well.

Dr. Turck:
Well with those best practices in mind, I want to thank my guests, Drs. Brian Smith and Charlie Strange, for joining me to discuss collaborative strategies for detecting and treating alpha-1 antitrypsin deficiency in patients with COPD. Dr. Smith, Dr. Strange, it was great having you both on the program.

Dr. Strange:

Thank you for having me.

Dr. Smith:
Happy to join you guys.

Announcer:
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