

Transcript Details

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ReachMD

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

Uncovering the Undiagnosed: Perspectives on screening for alpha1-antitrypsin (AAT) deficiency

Announcer

You're listening to ReachMD. This medical industry feature, titled Uncovering the Undiagnosed: Perspectives on Screening for Alpha-1 antitrypsin (AAT) deficiency, is sponsored by Grifols.

Jim

Thank you very much for the introduction today, Lori. And I'm going to now just briefly introduce the panelists who'll be talking with me today on the particularly emphasizing the questions on screening for alpha and antitrypsin deficiency. So three very engaged young folks who will be participating today. The first Monica Goldklang is from Columbia University in New York and she's a very active investigator in Alpha-1, as well as a practitioner. Secondly, we have Igor from UCLA who's engaged, not only in Alpha-1 education, but also is an intensivist and an expert in liver disease and the liver component of Alpha-1 antitrypsin. And lastly, Kyle Hogarth, who is a pulmonologist critical care specialist from the University of Chicago where he also is very well known for his interest in Alpha-1 antitrypsin and his ability to educate healthcare practitioners on this subject.

So we look forward to having a very engaging discussion focused on the eight questions today with these three panelists. The first question we're going to have today is a really important one and I have worked many years with rare lung diseases and it really comes down to the medical community knowing about Alpha-1 antitrypsin deficiency since Dr. Erickson and his colleagues described it more than 50 years ago. Yet it is estimated that approximately 90% of people with this genetic condition remained undiagnosed. Why do you think that is Kyle? What is your feeling about that problem that remains a big issue?

Kyle Hogarth

Jim, thanks for having me and you're right, it's an ongoing problem. And as I always tell people, because it's just rarely tested for, the problem with Alpha-1 is because it principally causes emphysema and COPD and chronic obstructed airways diseases, we see those day in and day out. And so I've often said Alpha's sort of lost in the noise of all of the obstructed diseases that all of us deal with. And so you referenced obviously the original discovery of Alpha-1 and that would probably represent the lowest hanging fruit. And what I mean by that is, do you really think we needed all this medical education to recognize a 40 year old end stage emphysema patient and a barely smoker? That doesn't make any sense. You know something's weird, right? And so that's fine, but as we all know, Alpha-1 has a wide variety of presentations.

And so getting away from the clinical diagnosis of Alpha-1, because there is no way to clinically diagnose this, to what it really is, which is a laboratory diagnosis, and the recognition that the suspicion for the disease comes with the diagnosis of an obstructed lung disorder and shifting your brain from ruling it in to ruling it out. And I always use the analogy, I've said ordered a lot of CT scans to rule out PE, and it's a big number and there's about this many PEs I have found.

And I haven't stopped ordering it once I'm suspicious of it. Well, if I'm passing out an inhaler for the diagnosis of COPD, then I'm going to test you for Alpha-1 and look, I want it to be negative, that's the other mindset we have. I don't want anyone to have this whole horrible disease. And so I'm really glad when it comes back negative. Man, when that test shows up anything, is it unbelievably important and I know my panelists and colleagues are going to talk about the other issues with the high carrier rate in the United States. We're going to see this disease continue on if we don't keep looking for it and being able to manage it. So I got a little long winded, so I'll shut up, but thanks.

Jim

That's really some very, very good insight. Monica, I'd love to hear your thoughts on that question also on why this orphan disease,

because there's less than 200,000, and as Kyle pointed out, we've been hearing about it for a good while, but why do you think people are not being diagnosed? And that's still a problem, I think even to this day. So what is your insight on that Monica?

Monica Goldklang

For me, I think that I agree with everything that Dr. Hogarth said. I do believe that this is something that there are certain people that it's very obvious, but part of it is about the medical education that we all received. And there's so much to think about in the differential diagnosis of this disease or that disease and if you start thinking about zebras very early in your training, you can become overwhelmed. And so I think that we've all been taught to kind of really not think about these zebras, but in fact it is a laboratory diagnosis and it's something that we all need to be thinking about.

But I think that people believe through our medical training that we know it when we see it, but as somebody who practices in New York City where there's a lot of population mixing and a lot of people of different ethnicities, I still diagnose easy Alpha-1 patients. And so it isn't just the patient that you can see it and that they're of Irish descent, we need to look at it in all of our patients and getting past that is something about us also as a community educating practitioners to understand there are treatments for it because I often hear people say, well, why would you test for it? It's a genetic disease. And so that's on us as a community to really educate.

Jim

Well, thank you both. I'm going to move on now to another very interesting question and that is what motivated you to become an Alpha-1 advocate and support systematic screening, which is what we're going to be talking for Alpha-1 antitrypsin deficiency. And I'd like to first pose the question to Dr. Igor, how did you start out there in UCLA on the west coast?

Igor Barjaktarevic

Sure. First, thanks Jim, and then thanks for having me in this great crowd. I actually didn't start with Alpha-1 on the west coast, I started on the east coast in New York. But that said, I'm a pulmonologist and interested in COPD. And this is actually the only genetic and very well described genetic cause of or familiar cause of COPD. But that said, I think the significance of this disorder goes much beyond the pulmonary medicine. It's a predisposition and it's a predisposition to hold set of various diseases that affect the respiratory tract or GI tract or skin, et cetera. So with that said, as a genetic disorder, actually this is in my mind a fairly low hanging fruit for any practitioner. This is a genetic disorder that has a very well described genotype and its relationship with the endotype and phenotype.

It is a genetic disorder that actually is very simple and very cheap and easy to test. And then the results that if you don't rule out as Kyle pointed out, but actually if, unfortunately you rule in, this is the piece of information that any physician, not pulmonology [inaudible 00:07:20] delivers and its very significant and has amazing impact on the life [inaudible 00:07:25] person independent on whether that person is sick and has significant lung disease because you are providing an explanation. This also actually significantly impacts the life of someone who does not have any evidence of disease of lung disease or gastrointestinal because lifestyle modification may make a major impact on the whole life lifetime of that patient. And then in addition, you do have impact on the family members and the relatives of a person who gets tested. So I think that pretty much the bang that you get for a buck actually is it's such significant that I think that anyone who tests contributes a lot.

Jim

Well, thank you, Igor. And let's move on to the next question. And we're going to put you back up Monica and then Dr. Big, Dr. Igor again. Monica, what criteria do you use within your practice to determine that someone is the appropriate to screen for Alpha-1 deficiency? How do you implement this in your practice? I'll start with you Monica, and then we'll go to Igor, okay?

Monica Goldklang

Yeah. So I'm very by the book, I screen everyone with diagnosis of COPD regardless of age of diagnosis, smoking exposure history, or their heritage. In addition, I screen everyone with asthma, with incomplete bronchodilator reversibility, and I believe that that's actually where we find a lot of these patients who are a little bit sick, but not very sick with Alpha-1. And in addition, I'm very blessed to have some wonderful colleagues within our hepatology clinic. And so every patient with cirrhosis, even if alcoholic cirrhosis is also screened for Alpha-1, and so we make a large number of diagnoses that way. Obviously as well, panniculitis, which I seem to only diagnose panniculitis in my alpha patients. It's very rare that a diagnosis of panniculitis backs me into Alpha-1 diagnosis just by nature of my practice.

Jim

That's really very interesting. Monica, as a background mentioned the great expertise in Alpha-1 at Columbia University and I, as a young guy, I did two months with Dr. Jerry Turino when he was at Columbia. And of course your colleague Janine D'Armiento is the president of the Alpha-1 Foundation. And you had great liver people, as you mentioned, David Brenner was there and many others. So that's a really a terrific background that you shared. And the [inaudible 00:09:56] Center for Alpha-1 that Dr. Turino had. Igor, how about you? How did it happen at UCLA with Dr. Tashkin and all those giants out there?

Igor Barjaktarevic

No, no, listen, this is a great question. And Monica really liked like nicely covered this. I mean, maybe what I can say is just like add or emphasize couple of things. So yes, there are things such as COPD and emphysema, but really adult onset asthma as Monica has mentioned, but also some common things that internal medicine we can see bronchiectasis. Bronchiectasis cell also should be a reason to screen. And then as you go beyond any end stage, we have a large end stage liver disease program, and we're a large liver transplant center. I see the most of patients who are evaluated for liver transplant. And I can tell you anecdotally in the past unclear etiology of liver failure was tested for Alpha-1 antitrypsin.

In addition, I would like to mention something that we occasionally also skip. My patients, their relatives get tested, their siblings, their parents. And then at the same time partners of people who are actually severe deficient actually get tested to get the projection for possible families, et cetera. And then I also need to mention one more thing. There is very particular new type of testing that is happening beyond our effort and it's like all these commercial genetic testing kits that are coming. And I'm sure all of us who deal with Alpha-1 are flooded with this healthy young people who just ring on the door of our offices looking for explanation, what the hell is this? I just did a test and I got a paper that says that I'm prone now to get lung and liver disease. So I think that independent of us, I think that with the availability of genetic testing and as we're moving forward this is going to become more and more topic about to talk about.

Jim

Those are excellent points that both of you made and like the liver, we had our what's called the Masec, the medical and science.

The liver, we had our what's called the MASAC, the Medical and Scientific Advisory Committee to the Alpha-1 board this past weekend. And I think 18 members are pulmonologists and only two are liver. And the real disease and liver of course, is childhood hepatology with the children's do seem to get it. So in terms of screening, it's just, it's relatively young disease to the pulmonary community in the sense that, Ron Crystal's studies and the registry, the historic registry, those studies go back not too many years ago, 20, 25 years ago. So it's pretty much new to a lot of pulmonologists, and we need to keep working to get to signal out. But particularly, as you point out that whole problem with the liver disease, I mean, that really is the seed of the disease is the liver and the need to be aware of it, and to get that into the awareness of the hepatologist, I think is a big challenge for all of us.

Kyle, we've been leaving you off the hook here too much, so I got to put you back to work. Is that okay?

Kyle Hogarth

Hmm. That's great. No problem.

Jim

Now we're going to take advantage of, you are really known and I know this from your work, a great communicator to the primary care. So knowing that most people with COPD are being cared for by a primary care provider, what do you say to those healthcare providers about the importance and ease of screening to rule out Alpha-1 antitrypsin deficiency? So we'll start with you because of your great experience.

Kyle Hogarth

Yeah. Well thank you. No, I appreciate that. Thanks for the compliment. So I think it's pretty straightforward here, and the reason I say that is, number one, primary care is set up to already draw blood. So if it's going to be tested for, from a blood draw, you're going to be drawing an annual CBC, or comprehensive panel, or thyroid scan, or lipids, whatever. So adding on one more blood test is straightforward. If blood's not an option, then obviously there are now buccal swabs as well, that are part of the being able to be tested, so there's a plenty of resources available.

But I think what it really comes down to is when it comes to lung disease. I agree with you 100%, majority of the COPD is being managed in primary care as it should be. And as part of that management and part of that evaluation, it's doing also the workup for the disease, and doing again, that rule out, and this is probably why it matters even more because nobody just suddenly has advanced, end stage lung disease and ends up in the four of our clinics, right? They got there over time. And so long before you're arriving to see me gasping and on oxygen, et cetera, you were progressing, and you saw somebody who diagnosed your COPD 15 years ago. And if they had diagnosed your Alpha-1 15 years ago, a lot more could have been done about it. And I think that's again where this information comes in. The ultimate in primary care would be to make this diagnosis before something becomes a major problem. We know, that's why we do screening for malignancies; find them at early stage. So let's find Alpha-1 or at least the lung disease from Alpha-1 at early stages.

I think the other key thing too, because there's a lot of drug development happening on the liver side. Right now, we don't have anything for the liver. I'm fairly optimistic as to what's coming down the pipeline. So if we're going to be drawing liver function tests and my two colleagues highlighted real clearly here that it's not just a pediatric disease. And so, when you have those abnormal liver function tests,

even in your alcohol user, let's work it up. So, I think if you're going to prescribe an inhaler, there's a reason, and there's obstructed disease, and finish the workup and it's that easy. There really isn't a lot of work either, right? I mean tacking on one more blood draw or a simple buccal swab. Oh, and by the way, we all have data that shows, once you explain to someone they're increased for lung disease related to their genetics, you're more likely to get them to quit smoking, which that is the ultimate primary.

Jim

Right, absolutely. Those are great points. Now I'd like to follow up a little bit. I have two questions for you to focus on as part B of this question, first is talk to me about a screening asthmatics and then secondly, can we talk about the Grifol screening program?

Igor Barjaktarevic

Sure. So, first of all agree with everything that Kyle said. In general, the data that we like to quote for a while is that a patient with Alpha-1 diagnosed with Alpha-1 on average he's previously like spent seven years looking for the , and seen at least three physicians. And we like to kind of talk about this data and you could argue that nowadays with the guidelines that are out there since 2003. And I think this is very that I remind it's 20 years, almost, that we have very clear indication to screen that there are no insurance issues with screening, and then they also industry facilitated the screening, which gets me to what you mentioned.

There are multiple industrial partners that work together along with Alpha-1 foundation, they all work together in an organized fashion in which these kids, the simple kids are kind of prepared, distributed to primary care to pulmonary offices. They are very simple. They can either be the finger sticks or even buccal swabs that they are doing; the simple, reliable methods of screening. I think, the relationship between industry and academic medicine in this is very fair and square industry is making the product. They are looking for their potential customers, but in order, that's for the greater good, that's for identification recognition of the disease.

And that gets back to what Kyle so has mentioned; when I do a blood work ice screen for Alpha-1. And I think that the provider should be familiar in their institutions. There might be several different tests and the Alpha-1 levels per [inaudible 00:18:08] may not be sufficiently good for screen. There are Alpha-1 phenotype or Alpha-1 genotype, and if there is a blood work, these test are readily available. If this comes just an add-on, it's a free screening. And I think that the way to really look at this is that this should be incorporated in a practice. In my COPD clinic when subject comes to be seen, actually, this is part of the workup. And in that sense, there is no reason for me to think of who should be screened; everybody gets screened and then we discuss exceptions not to screen. And I think that should be the appropriate-

Jim

Let me just ask you guys and anybody can chime in here, what do you think about the additional alleles and the buccal swab that we're picking up? Has that been of value to you? It has been to me because I guess I see a lot of very peculiar cases of Alpha-1, just because I've been in doing it for so long and finding those other alleles and working them up have been very interesting, and have led to some importance, the patients having their treatment, Walter. Has that been true of others, or is it too new? The screening with the buccal? Monica, could you-

Monica Goldklang

Yeah, Jim, I can give you a great story of a patient who had a phenotype on and she thought that she was MZ and that she had her child genotyped and the child came back actually as a MF. And as you know, the F allele, it is not functional, but on an isoelectric would look like an M. And so, this patient actually had walked around and she herself was a FZ, and so is functionally like a ZZ patient. And so, having something where we have a true genotype can only help us to make more diagnoses, and make precise diagnoses for our patients.

Kyle Hogarth

Yeah. I mean, Jim, it's a genetic disease. We want to know your genetics. And so the expanded genetic capabilities of the buccal swab test that that Grifols provides is a huge benefit. For me, it's all about clinic efficiencies; if your clinic is not to be able to use the buccal swab for whatever reason, then fine. If you're going to be seeing this patient, and drawing any amount of blood and you've made an obstructed lung disease diagnosis, tack on the Alpha-1 genotype, some labs call it a phenotype, but just order the genotype, so that it's coming with your next blood draw as well. Whichever way. In the end, when you've done the test you've done your duty as a physician to manage the disorder, the COPD that you diagnosed them with.

Jim

Sounds very good. That's very helpful. The next question; what has been the biggest value to you and your patients since you started screening COPD patients for Alpha-1? And this is, first I'd like to take a shot at, because I was here doing COPD even 50 years ago, that's how far back I go in the early '70s. But anyway, we didn't have screened earlier on, we send a level down and what have you. But it's been tremendous, not only for the individual patient, the fact that you get really accurate information, that's scientific that you can

really, because it is so accurate, hang your hat on it. And then, you can use that information to expand your search to the family members and use that as a great opportunity teaching point. And as many of you made, when we did the study a few years ago under I think it was the foundation where we screened everyone who was obstructed in the COPD in the pulmonary function lab, one in 10 were carrying that gene. I think, somebody said that before were carriers, very few disease, but a lot carriers. That's really, really, really important. So Kyle, I'm going to ask you to follow up to that; what is the risk of not screening for COPD? What kind of damage could be done?

Kyle Hogarth

That's a great question because let's just start. I think some of our colleagues have said of this already; let's just begin with the knowledge that there's a genetic predisposition for the development of are in lung disease that runs in your family. That automatically gives you the ability as both a clinician to talk with your patient, but then to provide the insight to the patient as to why some lifestyle modifications immediately matter. From either smoking or career choices, and then, obviously weight and alcohol intake, et cetera. So upfront, you can make changes, and they're not just a, "Oh sure, every doctor tells me to lose weight and not smoke." I can say, "No, definitively you, not anyone else, you genetically have to do this. And here's why. Here's what's going to happen." So right up front, that information matters.

But also, think about it from just put it into the mental context of any other disorder that you're not managing, and yet there it is continuing on and ravaging two separate organs, potentially. That's why this matters so much; because it allows us to manage the disorder. We have treatment for Alpha-1, especially when it's related to the lungs. We have a bunch under development in relation to the liver, but at the same time, let's go a different step; I diagnosed you with Alpha-1, let's say you actually at the moment have zero clinical manifestations other than some mild abnormal liver function tests. But now I'm about to put you on a statin. Maybe I'm going to rethink my medical approach to your other disorders, thinking upfront of potential damage to your liver. I don't know, but an informed patient and an informed clinician, make all the difference in regards to managing everything that we're dealing with in a primary care setting.

Jim

That's great. Thank you, Kyle. And I'd like to now put Monica and Igor on the firing line again. Now the first question is, how has the COVID-19 pandemic-

How has the COVID 19 pandemic and the need to consider underlying risk factors impacted your view of the importance of identifying patients that have Alpha-1? So Monica first, and then Igor. We've talked a little bit about this. And like I said, I learned more about COVID in the ICU from Monica. I didn't know anything about it when we were on a conference call one day. So Monica, I'll let you start off and then follow up with Igor. Okay?

Monica Goldklang

Yeah. So unfortunately in New York city, we had a really intense surge of COVID 19. And I was very lucky that, and I think all of us as a community were lucky that the Alpha-1 foundation, the messaging to our patients was very, very informative early on about the importance of just isolating, staying at home until we understood more. And as we spent time understanding more, we understood that actually the patients with alpha who did contract COVID 19 early in the pandemic, that some of those patients became very sick, some passed away. And through a whole body of research, it has turned out that at least in part, some of that is due to the way that Alpha-1 antitrypsin interacts with some of the enzymes that allow COVID 19 to enter the cells.

And so I think that now looking back, it makes sense why we've seen such severe disease in patients, but it's also been important for us as an Alpha-1 foundation CRC, to move forward in terms of educating our patients. Number one, about the importance of vaccine, but number two also about as society returns back to work, what are some of the work environments that are safe and potentially not safe for an Alpha-1 patient? And so we have really thought very hard about each individual patient and their return to work in a vaccinated setting and how risky or not risky that is for them based upon their Alpha-1 status.

Jim

Well, thank you very much. And as the panelists, and I know we've had a lot of this discussion about the risk to Alpha-1 patients. And fortunately data from alpha net that said that the majority of them stayed home. So the number of cases wasn't awful because we all thought the fatality would be high. But other studies show that, as Monica, and [Kyle 00:26:19], and Igor have been pointing out, there was very, very low, if any Alpha-1 in the lung during an event with COVID. Furthermore, with bundling, sometimes patients with Alpha-1, it's been a while since they received their last treatment, if they're on augmentation type of therapy. So Igor, do you have any perspective to add here on this question?

Igor Barjaktarevic

Yeah. I always like to add. And I know Monica really put it nicely, but maybe it's important to emphasize it really COVID impacted like multiple levels. And there is definitely this medical side of it. And there is very like technical and epidemiological side. So to add to the

medical side that Monica has mentioned, we know that Alpha-1 antitrypsin has more than a role of just like neutralizing neutrophilic. We know that it has immunomodulatory effect. We know that it's a good phase reactant. I mean, Alpha-1 does have this nice soothing immunomodulatory effect. And very soon into COVID we realized that this is not just upper respiratory infection. This is systemic infection. This is systemic infection with very significant immunological kind of consequences, immunological storms, et cetera.

So in that sense, definitely this is the medical impact of COVID onto our population. And that said, the technical, now I'm getting really to back into my office and sitting in this chair, impact of COVID on epidemiological side, really brought less direct interaction with patients, less ability to really see, to test patients. A lot of visits have turned into video visits. A lot of infusions have been compromised during the COVID era and skipped because of it. So I would say that the impact of the pandemic reaches multiple levels, but as you pointed out, Alpha-1 antitrypsin patients are well organized, Alpha-1 Foundation is keeping them organized. They kind of protected themselves quite well. And we did not have many cases of people who actually ended up dying with COVID of Alpha-1 antitrypsin.

Jim

I'm going to follow up that point about the patient and the patient perspective. And how has the patient experience, perspective, and reaction been regarding screening for Alpha-1? I like to ask, as a genetic disease, hereditary disease, I'd like to ask Kyle his opinion on that. Again, he's had a lot of experience in talking to the patients. So, Kyle?

Kyle Hogarth

Yeah, it's interesting. The patients actually are, when you ultimately make this diagnosis, clearly there's the shock of being told this, and that's a natural understanding. But it's also interesting that there's a sense of relief in a way, because inevitably, if there's been smoking involved, society is not very kind to smokers and lung disease, as we all know. And there's a sense of burden relieved actually, by kind of explaining that I had a genetic predisposition to this. And so I find frequently that the genetic aspect of this disease provides some level of sort of mental relief for some of my patients. Now, where we run into a lot of problems, and I spend a lot of time dealing with, and I'm sure my colleagues as well too, is then the extended family testing sometimes falls short.

And there is a frustrating, I want to put my head in the sand approach that some family members take, even with the act study through the foundation and the ability to get confidential testing and to get it not even necessarily in the doctor's office, to get around some of that. It's been frustrating, but over and over and over, I think we've been using some case examples, occasionally. I had a young Alpha-1 patient, was diagnosed in childhood as a double Z. And she called me one day and said, Hey, we're thinking about having kids. Is this going to affect my pregnancy? And she had no liver or lung disease clinically. And I said no, normal pregnancy, et cetera, et cetera. But get your spouse tested. And she said, oh, there's no lung disease in his family. Everybody in his family has heart disease. I said that's great, get him tested. Sure enough, he was a carrier.

And it really does highlight this very high carrier rate. And of course it doesn't change anything about them having children. It just made an informed decision, and especially on the monitoring of the newborn, especially obviously if there's jaundice. And so, when Igor talked about earlier about some of the sort of lay level blood testing, that's going on to test your ancestry and your heritage, I think when genetics was first coming out as a field and as a concept, there was understandably broad concern about my genetic information. But it really strikes me, especially amongst a younger generation, how open everybody is about genetics and how interested everyone is in genetics. And it's, I think comes again to say, look, there was a predisposition to this lung or liver disease you have. We want to be able to do something about that. And testing for it is so simple and so easy, and especially for the extended family, so that people again can make informed choices.

Jim

Thanks, Kyle. Monica, do you have any insight into this, the patient perspective?

Monica Goldklang

So thinking about a different part of the patient perspective has been all of these patients that have been diagnosed with Alpha-1 during the course of the pandemic and how hard their care has been, I've felt very fortunate that in this setting that we have been able to bring many patients in through telehealth, and that we were able to see people quicker and maybe earlier than necessarily when they would've felt comfortable coming into Midtown Manhattan to see us. But at the same time, there's something about being able to look somebody in the eye and really talk to them about the diagnosis, that it's virtually impossible to do via telehealth. And so we've tried to kind of come up with some hybrid ways of at least starting virtual, then catching them in house for PFTs or whatever else. But undoubtedly the care for patients during the pandemic with this sort of real surprise dropped in their lap has not been easy and has been something that hopefully we can all move forward from soon.

Jim

Thank you, Monica and Kyle.

Kyle Hogarth

Can I make one small plug again?

Jim

Yeah, please, please.

Kyle Hogarth

From a perspective of being a primary care physician, and I'll watch the nodding of the heads of my colleagues, taking care of Alpha-1 patients is a joy. They are an unbelievably informed and organized group on average. How often are you dealing with patients when you say, what inhalers are you on? And they go, I don't know, it's purple I think, maybe. But your Alpha-1 patients, they'll be the ones who'll come to you and say, Hey, did you see that article about the new blah, blah, blah? And you go, no, when did it come out? Oh, like 10 minutes ago. It's all over the web. The whole community's lit up about it. It is a wonderful thing to take care of these patients because they really are an informed and organized group. So go looking for them, and you too can join the club.

Jim

That brings you back to the old adage that Alpha's disease, the benefit that they received, was increase intelligence and creativity. Just a quick anecdote, one of the patient perspectives that I have, because there's a point here. I had a patient one day whose daughter I taught physical diagnosis. And he transferred his care to me late in life. And it was back in the old days. I threw up the x-ray and he had panlobular emphysema on the chest x-ray. Anyway, a long story, he's a genetic epidemiologist and he became passionate about studying the disease. And one of the things I was always working under the assumption, this is a Viking disease like CF. So it's a Caucasian disease. No, he went and looked at the African data. And of course the Spanish with the S gene that they get.

And sure enough, you should scream African Americans and Hispanics too. Please don't just scream Viking obvious. I thought that was pretty cool that with him not knowing he had it until late in life and then getting fired up. But we got to end this quickly because I'm using up too much time. But anyway, the last question here is to Dr. B. First Igor, what advice would you offer fellow HCPs, practitioners, healthcare practitioners that are looking to do more to diagnose and treat Alpha-1? Do you have any advice for them, how they can learn more about it? What do you tell them when they, other than writing an excellent consult note back to them? What are other things?

Igor Barjaktarevic

The message thing, Jim, like the message is very clear. Test for alpha one antitrypsin. And one thing is that first, it's around. We estimate a hundred thousand, maybe [inaudible 00:35:35] disease. But actually when you look at the carriers, we're talking about over 5 million in the United States. So if you saw COPD, if you have patients with asthma and COPD, probably do have some people who are carriers or severely deficient. Secondly, really this testing, it belongs to healthcare practitioners, because this is multi-systemic potential disorder. It's not all pulmonary, it is not all liver disease. So with that testing, you are covering actually multiple specialties. Dermatology.

With that testing, you are covering actually multiple specialties, dermatology, pulmonary, liver, GI, et cetera. So one stone hits a few birds. Then the testing is easy. The kits are available. Anybody can do it. Can be you, it can be your RN, your MA, it can be even the patient, him or herself, can actually participate the testing and it's free. And then finally, this is really a genetic disorder. It's a low hanging fruit to get into the what future is. And we're hopefully getting towards personalized medicine. This is one simple, easy step to get into that field, to be able to find genetic disorder, and not to have devastating news, but really to have impactful news to your patients. Sharing the finally positive diagnosis with pulmonologists is really sharing that patient still belongs to healthcare practitioner.

Jim

Kyle, what do you have? Then I'm going to ask Monica. Do you have any advice also where both of you in your interactions with the primary care HCP how to get better educated if they want to? Because some, I think you mentioned, have really become impassioned about this condition to wrap up.

Kyle Hogarth

Yeah, let's face it. On one level, every single one of us went into healthcare, no matter how you're practicing and what you do, because you want to make a difference. You want to change people's lives for the better. And I can't think of a better way than this because a simple test can dramatically shift how someone's lung disease and/or liver disease, or as Igor said this, the multi-systemic disease, is going to be managed and it's going to be managed proactively instead of reactively. And I thought that was always the basic tenet of all of medicine, but especially primary care. Let's be proactive. Let's find things before they become big problems.

And this simple thing allows you to become this essentially leader in lung health within your practice. There's a colleague who's a physician assistant in Georgia who took this on as just a passion project and the single greatest success was simply integrating it into a protocol within this large group practice to where patients who had abnormal spirometries, or came in with a diagnosis of COPD, got tested automatically right up front as part of the visit before they'd even seen the healthcare provider. And this person took it on as a

passion project, and of course, has made massive changes in multiple patients lives. A click of an order in an electronic record or a simple buccal swab that the patient can practically do themselves right there in clinic. Something so simple to make a massive impact, we can all be so lucky.

Monica Goldklang

I entirely agree and I would add on to all of those very on point comments that actually the future for Alpha-1 is bright. And I think that as doctors, we also there's a tendency to want to shield your patients from things that may be sad to them or may be harmful or may have implications for life insurance or other things. But the future for Alpha-1 is bright. It is not a death sentence. This is something that I really try to emphasize as well of what everybody else has said that in addition, when you find an alpha, they become a part of the alpha family, and there's probably no better patient group to work with than alphas. But it is a bright future I hope for them.

Jim

Thank you, Monica. I want to thank the panelists for this excellent discussion focused on these eight questions today. And we're obviously very passionate, but as the speakers have mentioned, this is a, although a rare disease and an orphan disease, the frequency of particularly the carrier state is quite high. So your screening will not be unproductive. It'll often be productive, particularly if you follow the guidance. If you know of them for early liver disease, early emphysema disease, familial disease, asthma that it's irreversible component, those kinds of things, and you'll be greatly rewarded. I think that's the thing I want to talk about most and hope you understand. The screening leads to positive results, particularly in the lives of our patients and their loved ones. And so as the three young panelists said today, it is extremely rewarding. So I have a question for the panelists. Monica, let me start with you. What are some resources you would recommend to HCPs to help them to learn more about screening and about Alpha-1 antitrypsin deficiency?

Monica Goldklang

So I recommend to all of the PCPs that refer patients to us that also that the Alpha-1 foundation is a very rich resource with regards to information for our patients and for their families. In addition to some of the educational material that can be found on the foundation's website, I found that the foundation is incredibly helpful with regards to talking to patients about insurance implications, helping them navigate through the whole Medicare, Medigap for proper coverage for infusions. And also our staunch advocates for making sure that there's familial screening and have put a ton of time and effort into making sure that familial screening can be done even in a confidential manner through the act study. I refer the patients to Alpha-1 Foundation, but in addition, I think that one of the most important things that a primary care doctor can do for an alpha-1 patient is to make sure that at least once a year that you're getting these patients into an Alpha-1 Foundation Clinical Resource Center so that they can have proper lung and liver evaluations to make sure that not only are they receiving the care that they should have for alpha-1, but in addition, that they're hearing about research studies that they may be eligible for as a part of their alpha-1 diagnosis.

Jim

Kyle, I'm going to end up with you. Our colleagues in industry have really played a major part, not only in the education programs through the foundation, but also in AlphaNet, but also independently by providing the screening kits and a place to have the screening test done well and very accurately. Do you have some insight onto that you would like to... what do you share, because you told me you did a lot of talking with the primary care doctors. What do you share with them about the screening and the availability of it and how they can learn?

Kyle Hogarth

Absolutely. So this is one of those wonderful areas where there is no shortage of resources. As everyone has said, starting with the foundation, and then also from a patient's perspective with the AlphaNet. And then obviously, this is one of these great scenarios where our industry partners play a huge role. When we've talked about the buccal swab tests, it's been mentioned how that's a free test. That free test is only available to you because of the commitment that a company like [inaudible 00:43:16] has made to this disease state and to this patient population. And when we say it's free, it completely free. And so that is an unbelievable commitment and it's all about finding these patients.

And then as was stated by both my colleagues, the Alpha Foundation, if ever there was a more amazing organization that's really designed to constantly fight for only one thing, which is to get rid of this disease, and then until we can do that day, manage it, and find all the patients. There's just an unbelievable amount of resources. So between the foundation, the AlphaNet, and the representatives of the company, you have more resources than you could ever hope for.

Jim

Well, thank you all for your insights. And we look forward to getting some feedback from the practitioners themselves on this subject. I'd like to thank the panelists for their engaging and enthusiastic discussion today. Also for all the participants, for listening to the program, I hope that you got a lot out of it, know more now about screening. I'd like to turn it over to the live portion of the program now.

Announcer

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