Knowledge of Alpha-1 Antitrypsin Deficiency Among Internal Medicine House Officers and Respiratory Therapists: Results of a Survey

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BACKGROUND: Alpha-1 antitrypsin deficiency is a common genetic condition that predisposes to emphysema and liver disease. Alpha-1 antitrypsin deficiency is under-recognized, so affected individuals often experience long delays in diagnosis and visits to multiple physicians before correct diagnosis. Reasoning that inadequate knowledge about alpha-1 antitrypsin deficiency could contribute to this under-recognition, we designed this study to evaluate internal medicine house officers' and respiratory therapists' (RTs) knowledge of alpha-1 antitrypsin deficiency. METHODS: We evaluated knowledge of alpha-1 antitrypsin deficiency with a Web-based test containing 30 multiple-choice questions. Invitations to take the test were sent via e-mail to all internal medicine house officers and RTs at The Cleveland Clinic main campus hospital. We assessed test scores by profession, years of training/experience, and self-assessed knowledge of alpha-1 antitrypsin deficiency. RESULTS: Of 332 invitees, 202 (61%) responded, of whom 165 (50%) provided complete responses (99 RTs, 66 physicians). The mean scores (percent of correct answers) were 54% and 52% for physicians and RTs, respectively ($P = .25$). The scores did not differ among the physicians when examined by subspecialty (pulmonary/critical care vs other) or post-graduate education level ($P = .94$). RTs who had graduated from a 4-year respiratory therapy program had a higher mean score than those who had graduated from a 2-year program (56% vs 50%, $P = .02$). Respondents' whose self-assessment of their knowledge about alpha-1 antitrypsin deficiency was "somewhat knowledgeable" had higher test scores than any other self-assessed knowledge level, regardless of profession. CONCLUSIONS: These results indicate a generally low level of knowledge about alpha-1 antitrypsin deficiency among physicians and RTs. Causes of under-recognition of alpha-1 antitrypsin deficiency, including the possibility of poor knowledge as a contributor, warrant further study. *Key words: alpha-1 antitrypsin deficiency; diagnosis; education; house officer; respiratory therapist.* [Respir Care 2010;55(3):322–327. © 2010 Daedalus Enterprises]

Introduction

Alpha-1 antitrypsin deficiency is a common but under-recognized condition.[1-5] Population-based screening studies suggest a United States prevalence of 1/3,000 births, and alpha-1 antitrypsin deficiency has been detected in approximately 3% of patients with chronic obstructive pulmonary disease.[6] Despite this frequency, ample evidence suggests that alpha-1-antitrypsin-deficient individuals often visit several physicians before the condition is identified and experience long diagnostic delays (mean 5−7 years) between symptom onset and correct diagnosis.[1-6]

Given that only a minority of the estimated 100,000 Americans with severe alpha-1 antitrypsin deficiency have

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been recognized,[1,5,6] efforts to enhance recognition have included the publication of official management guidelines by professional societies,[1] dissemination of testing opportunities (including free finger-stick test kits and confidential home-testing opportunities), and extensive continuing education offerings for pulmonologists,[1] primary care physicians,[7] and respiratory therapists (RTs).[8] Though many factors may contribute to under-recognition, suspecting and diagnosing alpha-1 antitrypsin deficiency require ample knowledge about its clinical presentation and features. We believe well informed primary care physicians and RTs would appropriately screen for the disease so that affected individuals would be diagnosed earlier.

The current study was undertaken to evaluate internal medicine house officers' and RTs' knowledge about alpha-1 antitrypsin deficiency and to determine whether understanding of alpha-1 antitrypsin deficiency increases over the course of training in internal medicine and/or training and experience as an RT. To do this, we evaluated the frequency of correct responses on a Web-based tool that asked for the participant's training and experience and self-assessment of his or her knowledge of alpha-1 antitrypsin deficiency, and tested this knowledge with 30 questions about alpha-1 antitrypsin deficiency. We then studied the relationships between the respondents' self-assessment ratings, test scores, and training and (for RTs) experience.

Methods

The study was approved by the institutional review board of The Cleveland Clinic.

The questionnaire was an abbreviated version of a comprehensive 75-question multiple-choice test that was initially developed (with funding from the Alpha-1 Foundation, Miami, Florida) to test RTs' knowledge of alpha-1 antitrypsin deficiency. The full test covers 9 categories: genetics, physiology, cell biology, liver, imaging, patient experience, epidemiology, clinical presentation, and treatment. For the present study, from the original questionnaire we randomly selected questions from all 9 categories to create a shorter 30-question questionnaire, on the assumption that a shorter questionnaire would have a higher response rate. We added 5 demographic questions about training, experience, and (for physicians) subspecialty. A cluster of 10 questions regarding clinical recognition and diagnosis of alpha-1 antitrypsin deficiency was identified among the final 30 questions.

Invitations to take the test were sent via e-mail to all internal medicine house officers ($n = 128$) and RTs ($n = 204$) at The Cleveland Clinic main campus hospital. Respondents clicked on a link to the Web-based survey and completed the test online. E-mail invitations were sent twice over a 6-week period. Respondents were offered a $10 gift certificate (for coffee) on completion of the test.

Physician and RT test performance was quantitated as a score (ie, percent of correct answers). Respondents were asked to self-assess their knowledge of alpha-1 antitrypsin deficiency, on a scale from "no practical knowledge" to "expert" by answering a 5-item multiple-choice question. We compared the test scores with Student's t test, Mann-Whitney rank sum test, chi-square test, or one-way analysis of variance (post hoc analysis with the Holm-Sidak method), as appropriate. P values $\leq .05$ were considered significant.

Results

A total of 202 invitees responded (202/332 = 61%). Thirty-seven records were deleted due to incomplete answers, so 165 complete responses composed the final study sample (165/332 = 50%). Sixty percent of the respondents were RTs. Table 1 presents the demographic features of the 165 respondents.

The range of percent-correct test responses was 23–77% for the physicians and 23–90% for the RTs. Overall, there was no statistically significant difference between the physician and RT groups (mean score 54% vs 52%, $P = .25$). Figure 1 shows that 50% of the physicians' scores were below 55% (ie, the median score), whereas the RTs' median score was 50% correct.

Also, Figure 1 allows easy appreciation of how many individuals in each group scored identically; the rows of horizontal clustered dots indicates identical scores. Thus, for example, Figure 1 shows that there were identical scores

Table 1. Demographic Features of Respondents

Internal Medicine House Officers	$n = 66$	(%)
Postgraduate year		
1	20	(30)
2	26	(40)
3	20	(30)
Specialty		
Pulmonary/critical care	6	(9)
Other	15	(23)
None indicated	45	(68)
Respiratory Therapists	**$n = 99$**	**(%)***
Years of professional education		
2	68	(69)
4	25	(25)
Incomplete response	6	(5)
Years of clinical experience		
< 1	12	(12)
>10	40	(40)
Incomplete response	47	(47)

* Percentages do not sum to 100 because of rounding.

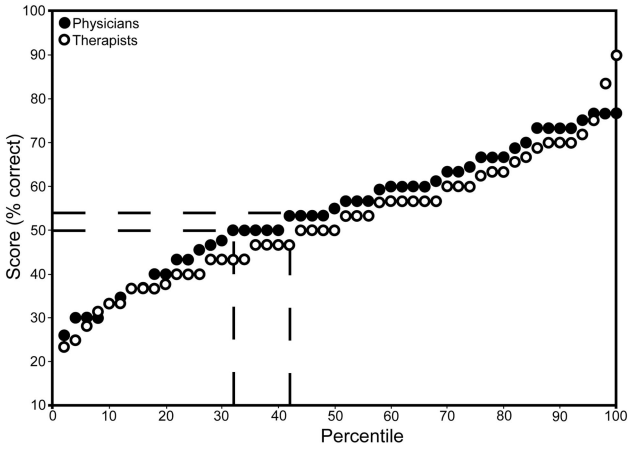

Fig. 1. Percentiles plot of percent-correct responses for internal medicine house officers and respiratory therapists, showing the percent of observed scores that is less than a particular score value. The dashed lines indicate that 41% of the scores were below 54 and 31% of the scores were below 50. A percentile is the value of a variable below which a certain percent of observations fall, so the 50th percentile is the value (or score in this case) below which 50% of the observations occur (the 50th percentile is equivalent to the median).

of 50% correct. The dashed lines indicate that 41% of scores were below 54% correct and 31% of the scores were below 50% correct. The absence of dots between scores 50% correct and 54% correct indicates there were no scores in that range. Therefore, by subtracting the 2 percentiles (ie, 41 –31) we see that 10% of the scores were identical values of 50%.

Table 2 presents the percent-correct responses by individual question (stratified by respondent type) by the 9 topic categories and by the 10-question cluster regarding recognition and diagnosis of alpha-1 antitrypsin deficiency. For the questions regarding clinical presentation, the range of percent of correct responses was 20–89% (mean 59%) for the internal medicine house officers, and 33–78% (mean 63%) for the RTs. For the 10-question cluster on clinical recognition and diagnosis, the range of percent of correct responses was 23–92% (mean 63%) for the physicians, and 30–92% (mean 61%) for the RTs. There were no significant differences in scores between the physicians and RTs in any question category.

There was no significant difference between the median scores of the 7 physicians whose declared specialty was pulmonary/critical care and the other physicians (53% vs 57%, $P = .36$ via Mann-Whitney rank sum test).

Figure 2 shows that there was no significant association between the test scores and self-assessment of alpha-1 antitrypsin deficiency knowledge. Overall, the mean overall scores differed among the self-assessed knowledge levels ($P < .001$), but pairwise comparisons showed that the mean test score for those who self-assessed as "somewhat

Table 2. Scores by Question and Category

Topic Category	Question	Correct Answers (%)		*P*
		Physicians	Respiratory Therapists	
Genetics	22	74	79	
	23	52	63	
	1	59	54	
	Mean	62	65	.77
Physiology	5	27	20	
	2	70	59	
	4	80	66	
	3	38	33	
	Mean	54	44	.20
Cell biology	6	52	45	
	21	26	30	
	7	73	73	
	Mean	50	49	> .99
Liver	10	86	92	
	8	70	68	
	9	62	28	
	Mean	73	63	.17
Imaging	12	23	28	
	11	92	92	
	Mean	58	60	.89
Patient experience	13	68	59	
	24	56	36	
	25	71	64	
	Mean	65	53	.11
Epidemiology	14	29	26	
	15	38	31	
	Mean	33	29	.65
Clinical presentation	16	71	74	
	18	89	78	
	17	20	33	
	19	58	68	
	Mean	59	63	.66
Treatment	26	27	20	
	30	12	15	
	27	30	27	
	28	35	36	
	20	58	54	
	29	83	89	
	Mean	41	40	> .99
Recognition and diagnosis	2	70	59	
	3	80	66	
	4	38	33	
	8	70	68	
	10	86	92	
	11	92	92	
	12	23	28	
	13	68	59	
	21	26	30	
	22	74	79	
	Mean	63	61	.88

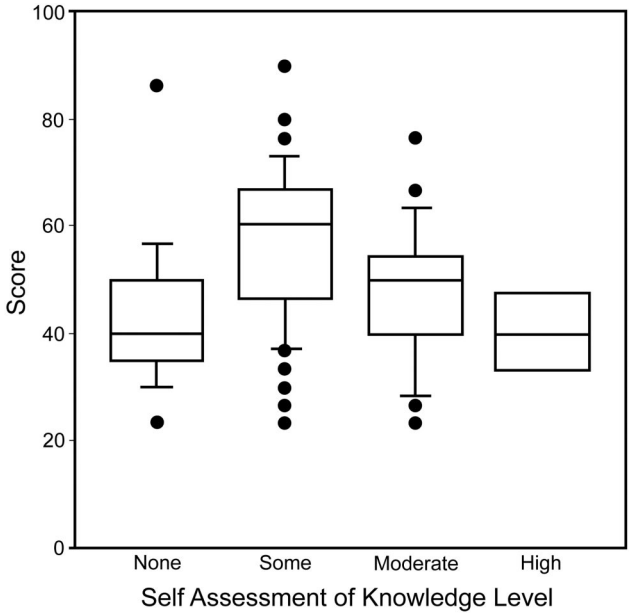

Fig. 2. Relationship between self-assessment of knowledge about alpha-1 antitrypsin deficiency and the percent of correct responses among internal medicine house officers and respiratory therapists. With each box plot, the lower boundary indicates the 25th percentile, the line within the box is the median, and the upper boundary indicates the 75th percentile. The error bars above and below the box indicate the 90th and 10th percentiles. The dots represent data points below the 25th percentile and above the 75th percentile. There was no significant relationship between self-assessment of knowledge about alpha-1 antitrypsin deficiency and the percentage of correct responses (combined data for the physicians and respiratory therapists; $r^2 = 0.002$, $P = .56$).

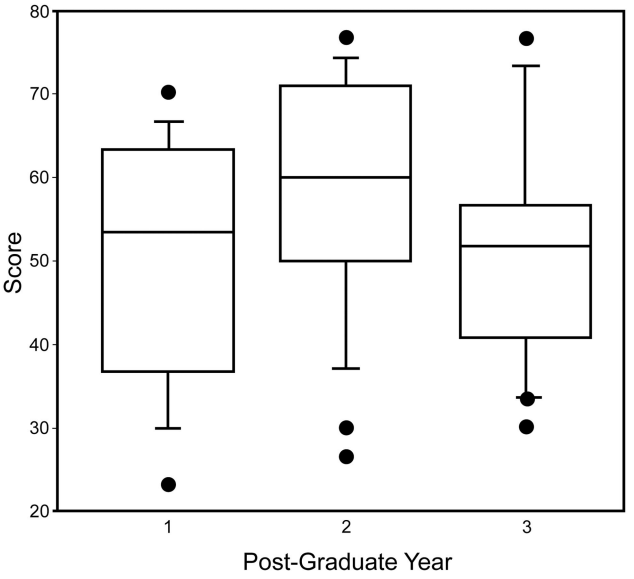

Fig. 3. Relationship between post-graduate year and percent of correct responses among internal medicine house officers. With each box plot, the lower boundary indicates the 25th percentile, the line within the box is the median, and the upper boundary indicates the 75th percentile. The error bars above and below the box indicate the 90th and 10th percentiles. The dots represent data points below the 25th percentile and above the 75th percentile. Physicians' test scores did not differ by post-graduate education level ($P = .94$).

knowledgeable" was greater than any of the other levels. None of the other pairwise comparisons showed significant differences. The result was similar when stratified by profession; those who self-assessed as "somewhat knowledgeable" had the highest test scores (physicians 60% correct, RTs 56% correct, $P < .001$). The physicians' test scores did not differ by post-graduate education level ($P = .94$) (Fig. 3), but the RTs who graduated from 4-year respiratory therapy programs had a higher mean score than those who graduated from 2-years program (56% correct vs 50% correct, $P = .02$) (Fig. 4).

Discussion

Our main findings are:

1. Based on test performance, knowledge of alpha-1 antitrypsin deficiency was generally low and did not significantly differ between the internal medicine house officers and the RTs.

2. Similarly, the house officers who declared a specialty interest in pulmonary/critical care did not have greater knowledge of alpha-1 antitrypsin deficiency. In neither group was self-assessed greater knowledge of alpha-1 antitrypsin deficiency associated with better test performance.

3. There was little evidence of increasing knowledge of alpha-1 antitrypsin deficiency by level of training among the physicians, but there was modestly better test performance by the RTs who had attended 4-year programs, versus 2-year programs. The findings suggest that, at least among the physicians in this sample, knowledge of alpha-1 antitrypsin deficiency did not accrue with experience, and, among the RTs the effect of more education on knowledge of alpha-1 antitrypsin deficiency was modest.

This study extends available knowledge by assessing internal medicine trainees' and RTs' knowledge of alpha-1 antitrypsin deficiency. Many reasons could cause clinicians to fail to recognize alpha-1 antitrypsin deficiency. For example, limited time for medical visits and the need to address more common conditions (eg, hypertension, diabetes) could cause primary care physicians to overlook alpha-1 antitrypsin deficiency as a cause of obstructive lung disease. Similarly, high work-load of RTs could cause alpha-1 antitrypsin deficiency to go unrecognized. Also, under-recognition of chronic obstructive pulmonary disease, perhaps related to the unavailability of spirometry measurements, would be expected to cause underlying alpha-1 antitrypsin deficiency to go unappreciated. Lack of conviction that diagnosing alpha-1 antitrypsin deficiency

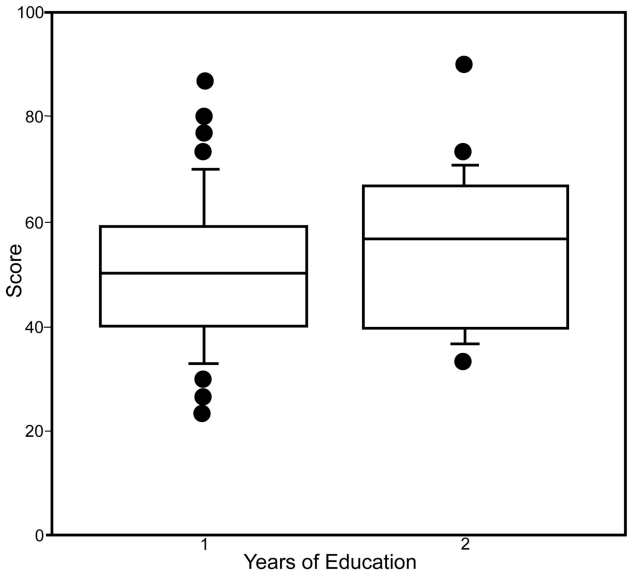

Fig. 4. Relationship between years of education and percentage of correct responses among respiratory therapists. With each box plot, the lower boundary indicates the 25th percentile, the line within the box is the median, and the upper boundary indicates the 75th percentile. The error bars above and below the box indicate the 90th and 10th percentiles. The dots represent data points below the 25th percentile and above the 75th percentile. Scores differed between the respiratory therapists with 2 years versus 4 years of formal respiratory therapy education ($P = .02$).

has treatment implications could lead clinicians to defer testing for the condition.

Finally, inadequate knowledge of the clinical presentation or features of alpha-1-antitrypsin-deficient individuals or failure to appreciate the frequency of the condition could cause clinicians to forego testing for alpha-1 antitrypsin deficiency, even in the face of official guidelines[1] that recommend testing all symptomatic adults with fixed air-flow obstruction on pulmonary function tests. Although our study does not address the causes of under-recognition of alpha-1 antitrypsin deficiency, our findings that internal medicine trainees and RTs had mean test scores of 54% and 52%, respectively, and mean scores of 63% and 61%, respectively, on the questions about clinical recognition and diagnosis of alpha-1 antitrypsin deficiency is consistent with the possibility that inadequate knowledge of alpha-1 antitrypsin deficiency could contribute to its under-recognition.

Our study also has implications regarding how to enhance clinicians' knowledge of alpha-1 antitrypsin deficiency. Specifically, these data generally challenge the idea that knowledge of alpha-1 antitrypsin deficiency increases with years of training in internal medicine or, to any striking extent, with years of respiratory therapy training. Recognizing the limitation of our cross-sectional study to address the longitudinal acquisition of information, the apparent lack of increasing knowledge of alpha-1 antitrypsin deficiency with experience and/or training suggests a need for formal continuing medical education. However, given the sometimes limited impact of formal continuing medical education on changing clinicians' practice,[9] our data invite consideration of novel ways to enhance clinicians' alpha-1 antitrypsin deficiency knowledge. The success of protocols executed by RTs[10-12] suggests that implementation of a protocol that prompts the RT to seek alpha-1 antitrypsin testing for patients with established air-flow obstruction might enhance detection. Current efforts to enhance diagnosis of alpha-1 antitrypsin deficiency by physicians and RTs include issuance of written prompts to test for alpha-1 antitrypsin deficiency when pulmonary function tests indicate obstruction,[13] issuance of physician alerts via the electronic medical records system, and empowering RTs to offer alpha-1 antitrypsin deficiency testing to patients at the time of pulmonary function testing if the results show obstruction. All of these approaches represent efforts to implement the "level A" recommendation by the American Thoracic Society, European Respiratory Society, American College of Chest Physicians, and American Association for Respiratory Care that symptomatic adults with obstruction should be tested for alpha-1 antitrypsin deficiency.[1]

Limitations

First, because the response rate was low, we cannot exclude the possibility of a sample bias that might cause our results to either overestimate or underestimate true knowledge of alpha-1 antitrypsin deficiency among internal medicine house officers and RTs. On the one hand, even though self-assessed knowledge correlated poorly with actual test performance, the test results in this study could be inflated if respondents were inclined to respond to showcase their knowledge. Conversely, those who considered themselves already knowledgeable may not have found the invitation interesting enough to respond, which might cause the observed rate to underestimate the actual knowledge level. Similarly, because the number of respondents in some categories (eg, the internal medicine house officers who declared a specialty interest in pulmonary/critical care) was small, the study's power to identify differences between compared groups is limited.

Second, as this is a single-institution study, the findings may not generalize to all internal medicine house officers and RTs. Regularly scheduled teaching for internal medicine house officers and RTs about alpha-1 antitrypsin deficiency, and a long history of research about alpha-1 antitrypsin deficiency at The Cleveland Clinic,[14,15] may cause performance at this institution to overestimate that at others with less experience with alpha-1 antitrypsin deficiency. However, in the context of our institutional experience, the relatively poor test performance we found in-

dicates the need for better methods of educating house officers and RTs to assure a high degree of awareness and knowledge of alpha-1 antitrypsin deficiency.

Finally, although the test was developed with the intent of testing clinically relevant aspects of alpha-1 antitrypsin deficiency, the test scores may not correlate well with the level of clinical management or recognition of individuals with alpha-1 antitrypsin deficiency. We emphasize that our results cannot be construed as explaining the well documented phenomenon of under-recognition of alpha-1 antitrypsin deficiency, and we offer the following example of a scenario in which early detection of an alpha-1-antitrypsin-deficient individual might occur despite the clinician's low knowledge of the condition. If, despite not suspecting the condition, the internal medicine house officer arranged early pulmonary consultation and the consultant tested for alpha-1 antitrypsin deficiency, prompt detection would occur despite initial lack of appreciation of alpha-1 antitrypsin as a possibility by the house officer.

Conclusions

Overall, our findings reinforce the need to enhance clinicians' awareness of and expertise regarding alpha-1 antitrypsin deficiency. Our hope is that these results will prompt continued investigation of strategies to cascade knowledge of alpha-1 antitrypsin deficiency through the communities of clinicians who may be seeing and caring for these individuals.

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