

2.15 Alpha-1 Antitrypsin Deficiency: Evaluating the Effectiveness of Health-Related Quality of Life Data in Tracking Disease Severity and Progression

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Background: Alpha-1 antitrypsin deficiency (AATD) is an inherited disorder that often causes lung disease, particularly COPD [1]. AATD significantly impacts health-related quality of life (HRQoL) [2]. While spirometry and radiology are commonly used to assess disease severity in AATD [3], HRQoL measures are often overlooked. This project aimed to compare objective clinical investigations with HRQoL measures in determining disease severity and progression.

Methods: Patients with confirmed ZZ, SZ and MZ phenotypes (n=516) enrolled in the national AATD registry were selected. A cross-sectional analysis and a longitudinal study were conducted, comparing demographics, forced expiratory volume in one second (FEV₁), carbon monoxide diffusing capacity (DLCO), and the St. George's Respiratory Questionnaire (SGRQ).

Results: SGRQ mean total scores were highest in ZZ patients for active, past and never smokers when compared to SZ and MZ (table 1). SGRQ scores significantly correlated with FEV₁% predicted ($r = -0.3272$, $p=0.0069$) and DLCO% predicted ($r = -0.4818$, $p<0.001$) (table 2). Patients receiving augmentation therapy have a mean SGRQ score change of -15.5 over a mean time of 7.39 years (figure 1).

Conclusion: Subjective HRQoL measures can complement objective clinical assessments in assessing lung disease severity in AATD.

Keywords: Alpha-1 antitrypsin deficiency (AATD), chronic obstructive pulmonary disease (COPD), health-related quality of life (HRQoL), augmentation therapy.

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Table 1. Most recent spirometry and QoL results for the 3 phenotypes classified by smoking history.

k	Active smokers			Past smokers			Never smokers			P-value
	ZZ	SZ	MZ	ZZ	SZ	MZ	ZZ	SZ	MZ	
n	9	9	22	134	54	81	85	55	67	
Pack-years, mean	16.82	20.92	49.38	20.2	24.34	26.24	n/a	n/a	n/a	0.1129
Most recent CAT score, mean	22	18	10	16.19	15.35	13.13	10.58	3.63	13.67	0.0175
Most recent mMRC score, mean	1.75	3	1	1.64	1.45	1	0.54	0.23	0.54	<0.0001
SGRQ total score, mean	47.75	12	n/a	32.13	28.18	10.75	20.95	10.13	8.25	0.021
Most recent FEV₁/FVC, mean	0.64	0.65	0.60	0.49	0.67	0.66	0.69	0.78	0.74	<0.0001

Most recent FEV₁ % predicted, mean	76.78	73.22	75.68	59.94	84.17	84.64	89.32	103.8	95.13	<0.0001
Most recent DLCO % predicted, mean	79.71	72	67.73	54.34	78.96	79.46	83.5	90.63	84.51	<0.0001

Table 2. Correlation analysis between SGRQ, subjective measures and PFTs.

Variables	Pearson correlation coefficient (compared with SGRQ)	p-value
Age, n=56	0.1027	0.4512
Pack-years, n=35	0.3582	0.0173
FEV₁% predicted, n=56	-0.3272	0.0069
DLCO% predicted, n=50	-0.4818	<0.0001
CAT scores, n=41	0.6049	<0.0001
mMRC scores, n=47	0.6417	<0.0001

Figure 1. Changes in SGRQ scores, DLCO% and FEV₁% predicted over a mean period of 7.39 years for patients receiving augmentation therapy.

