9.01 Evaluating Liver Disease in Alpha-1 Antitrypsin Deficiency (AATD)

Fiachra O'Meara, Simon Carty, Daniel Fraughen, Tomás Carroll, Hassaan Yousuf, Cedric Gunaratnam, John Ryan, Noel G. McElvaney.

Royal College of Surgeons Ireland

Alpha-1 Antitrypsin Deficiency (AATD) is the commonest genetic cause of COPD, while also increasing susceptibility to liver disease. This study aimed to examine the extent of liver disease in AATD patients in Ireland.This cross-sectional study included 169 patients from the Irish National AATD registry. "PiZZ", "PiSZ" and "PiMZ" genotypes were assessed using FibroScan; a transient elastography (TE) instrument for non-invasive evaluation of liver steatosis and fibrosis. TE's Controlled Attenuation Parameter (CAP [dB/m]) and Liver Stiffness Measurement (LSM [kPa]) assesses steatosis and fibrosis, respectively.32% of "PiZZ" patients showed LSM values ≥ 7.1kPa (stage 2 fibrosis) with an overall mean of 8.41kPa. In "PiSZ" and 'PiMZ" patients, 24% LSM values were ≥ 7.1kPa with a mean of 6.76kPa and 6.68kPa, respectively. All cohorts showed significant levels of steatosis with 71% of "PiMZ' and 60% of "PiSZ" patients yielding a CAP value ≥ 238 dB/m (stage 2 steatosis).

Liver steatosis and fibrosis is under-recognised in AATD. TE is a quick, quantitative, non-invasive imaging modality that is more sensitive than ultrasound in identifying steatosis and fibrosis and could be more readily used to facilitate the work-up of AATD patients and identify those with asymptomatic advancing liver disease.

Table 1(9.1). Characteristics of "PiZZ", "PiSZ" and "PiMZ" Phenotypes

	ZZ	SZ	MZ	All Phenotypes
Demographics				
n=	74	25	70	169
Male	34	10	34	78
Female	40	15	36	91
Age (Mean)	52.88 (17.14)	57.90 (11.58)	53.17 (15.19)	53.74 (15.64)
Height (cm) (mean±SD)	169.83 (9.05)	163.14 (9.03)	168.43 (9.55)	168.29 (9.43)
Weight (kg) (mean±SD)	80.19 (20.7)	74.37 (13.87)	79.10 (16.57	78.87 (18.64)
BMI (kg/m^2) (mean±SD)	27.76 (6.8)	28.09 (5.46)	27.92 (5.63)	27.86 (6.25)
FibroScan Finding				
CAP (dB/m)	253.78 (72.91)	258.84 (53.06)	276.96 (60.7)	264.13 (65.92)
n= S0 (<238)	33	10	20	63
n= S1 (238-260	6	4	3	13
n= S2 (260-290)	7	3	15	25
n= S3 (>290)	28	8	32	68
LSM (kPa)	8.41 (8.55)	6.76 (4.83)	6.68 (4.56)	7.45 (6.66)

n= F0-1 (<7)	50	19	53	122
n= F2 (7-11)	14	4	10	28
n= F3 (11-19)	4	1	5	10
n= F4 (>19)	6	1	2	9