

	ANI>0 (n = 18)	ANI<0 (n=67)	Odds ratio	p-value
Age (years)	62.2±9.4	61.8±9.8		0.97
Male	100% (18/18)	40.3% (27/67)	Infinity	<0.001
Body mass index	28.7±3.9	31.7±4.6		0.02
Arterial hypertension	61.1% (11/18)	59.7% (40/67)	1.06	0.86
Diabetes mellitus	50% (9/18)	40.3% (27/67)	1.48	0.64
Dyslipidemia	38.9% (7/18)	53.7% (36/67)	0.55	0.39
Recorded alcohol consumption:				
0 g/week	72.2% (13/18)	61.2% (41/67)	1.71	0.51
10-20 g/week	23.1% (3/13)	63.4% (26/41)	0.17	0.01
30-60 g/week	15.4% (2/13)	12.2% (5/41)	1.31	0.54
10-20/30 g/day (female/male)	7.7% (1/13)	4.9% (2/41)	1.65	0.57
	53.9% (7/13)	19.5% (8/41)	4.85	0.03
Fibrosis (Kpa)	7.6±3.6	5.9±1.7		0.18
CAP	292.7±46.6	305.6±51.9		0.41
AST (U/L)	46.7±35.7	34.1±15.9		0.08
ALT (U/L)	38.9±33.6	39.1±23.9		0.39

Comparison of ANI>0 and ANI<0 groups.

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P-14 DETECTION OF CIRRHOSIS DUE TO ALPHA-1 ANTITRYPSIN DEFICIENCY IN ADULTS: PHENOTYPE STUDY IN COSTA RICA

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Conflict of interest: No

Introduction and Objectives: Alpha-1 antitrypsin (A1AT) levels are normal in up to 20% of liver diseases, and this protein elevates in inflammatory states, causing false negatives. This disease does not follow an autosomal recessive inheritance pattern, so the classical concept of homozygosity does not apply. Instead, two codominant alleles manifest as liver or lung disease. *Objectives:* To determine the phenotypes associated with A1AT-related liver disease in Costa Rica.

Patients / Materials and Methods: Phenotypes detected in patients with suspected A1AT deficiency from 2014 to July 2024 were analyzed. Phenotype identification was carried out using iso-electric focusing in agarose gel with immunofixation. The presence of liver disease was determined through clinical, laboratory, and imaging findings.

Results and Discussion: During the specified period, 371 phenotype studies were conducted on 187 women and 184 men. The identified phenotypes were: 15 ZZ probands, 22 MZ probands, 1 SZ proband, 7 MS probands, 1 SS proband, 1 null proband, 1 M/null proband, 31 MM probands, and 2 null/null probands. No Z/null proband was detected. Among 53 probands, there were: 10 ZZ, 13 MZ, 1 SZ, 4 MS, 1 SS, 1 null, and 23 MM. It was established that the risk of liver disease is slightly increased in MZ, increased in SZ, and very increased in ZZ. Cirrhosis was diagnosed in 19 probands: 7 ZZ, 7 M/null, 4 MZ, and 1 SZ.

Conclusions: A1AT quantification has a 20% false-negative rate, so phenotype testing is recommended when there is suspicion. In Costa Rica, the ZZ variant has the highest risk of liver disease, followed by SZ and MZ; the M/null phenotype was also detected as a cause of liver disease. Medical monitoring is necessary, and in doubtful cases, genotype testing should be performed.

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P- 15 GENETIC AND CLINICAL CHARACTERISTICS IN LEAN MASLD PATIENTS WITH AND WITHOUT CIRRHOSIS IN LATINOAMERICAN POPULATION

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Conflict of interest: No

Introduction and Objectives: Metabolic dysfunction associated esteatotic liver disease (MASLD) is the most common chronic liver disease worldwide. It is associated with metabolic conditions and can also occur in lean patients with a normal or low BMI. Polymorphisms in PNPLA3, TM6SF2 and MBOAT7 genes have been linked to an increased risk of developing hepatocellular carcinoma (HCC) and greater severity of fibrosis. This study aimed to assess clinical and genetics characteristics in Latin American lean MASLD patients with and without cirrhosis.

Patients / Materials and Methods: This descriptive cross-sectional study evaluated 148 patients from an international database of chronic liver disease patients (ESCALON), including Colombia, Brazil, Chile, Ecuador, Argentina and Perú. MASLD was identified in patients with a BMI≤ 25. Patients with alcohol-associated and viral -related liver diseases, as well as other liver conditions, were excluded. Clinical features and main MASLD-related pathogenic variants were evaluated in this population. We used BlueSky software to evaluate two sample t -test for cirrhotic vs no cirrhotic variables. The assessment included the median age range and average BMI.

Results and Discussion: A total of 102 patients (69%) were found to have cirrhosis, with 57% being female and a median age of 65,6 years.42% had HCC, 39% had diabetes mellitus(DM), 14% had dyslipidemia, and 28% had hypertension (HTN).Common genetic variants were evaluated in 60.8% (90/148) of the study population with the following distribution: PNPLA3 (rs738409): 45,6% (GG), 43,3% (CG);MBOAT7(rs641738): 10%(TT), 43,3%(CT);TM6SF2(rs58542926): 0%(TT), 12,2%(CT) and 87,8%(CC);HSD17B13(rs72613567):1,2%(TT), 16,3%(AT);GCKR(rs1260326):33,3%(CC), 50%(CT).The characteristics by group and the differences found are shown in table 1

Conclusions: Cirrhotic patients were older, with higher rates of diabetes mellitus, hypertension, dyslipidemia and HCC. The PNPLA3 GG variant was predominant in cirrhotics compared to non-cirrhotic patients, with no significant differences between groups in the other variants