

CLINICAL PHENOTYPES OF ITALIAN AND SPANISH PATIENTS WITH ALPHA-1-ANTITRYPSIN DEFICIENCY

Authors: Barbara Piras¹, Ilaria Ferrarotti², Beatriz Lara³, Maria Teresa Martinez⁴, Ana Bustamante⁵, Stefania Ottaviani², Pietro Pirina¹, Maurizio Luisetti², Marc Miravittles⁶

Center:¹Institute of Respiratory Diseases, Sassari University - Italy

² Centro per la Diagnosi del Deficit Ereditario di Alfa1-antitripsina, Dipartimento di Medicina Molecolare, SC Pneumologia, Fondazione IRCCS Policlinico S. Matteo, Università di Pavia, Italy

³ Servei de Pneumologia, Hospital Universitari Arnau de Vilanova, Lleida, Spain.

⁴Servicio de Neumología. Hospital 12 de Octubre, Madrid, Spain

⁵Servicio de Neumología, Hospital Sierrallana, Torrelavega, Spain.

⁶Institut d'Investigacions Biomèdiques August Pi i Sunyer (IDIBAPS). Hospital Clínic. Ciber de Enfermedades Respiratorias (CIBERES). Barcelona, Spain.

Supplementary material

Table S1. Rare genotypes identified in the Italian and Spanish registries of individuals with AAT deficiency

Genotype	Italy	Spain	Total
Mmalton/Mmalton	5	1	6
Mprocida/Mprocida	2	0	2
I/Mprocida	1	0	1

Plowell/Q0mattawa	1	0	0
Q0/Q0	9	0	0
S/S	3	0	0
S/Mmalton	4	0	0
S/Plowell	2	0	0
S/Mheerlen	1	0	0
Z/Mmalton	10	3	13
Z/Mprocida	8	0	8
Z/I	5	1	6
Z/Mwurzburg	4	0	0
Z/Plowell	2	0	0
Z/Q0	3	0	0
Z/Mbrescia	1	0	0
Z/Mpalermo	0	2	2
Ybarcelona/Ybarcelona	0	1	1
Z/P	0	1	1
S/I	0	2	2
M/Q0bellingham	0	1	1
Total	61	12	73