Title: Health-related quality of life in emphysema due to alpha-1-antitrypsin deficiency

Dr. Sandra 2901 Manca mancasandra@tiscali.it MD ¹, Dr. Esther 2902 Rodriguez estherod@vhebron.net MD ², Dr. Arturo 2903 Huerta ahuerta@clinic.ub.es MD ³, Dr. Maria 2904 Torres mtordur@yahoo.es MD ⁴, Dr. Lázaro 2910 Lourdes lourlazar@gmail.com MD ⁵, Dr. Sergio 2911 Curi sergio.curi.chercoles@navarra.es MD ⁶, Prof. Pietro 2909 Pirina pirina@uniss.it MD ¹ and Dr. Marc 2905 Miravitlles marcm@separ.es MD ². ¹ Istituto Di Malattie Dell'Apparato Respiratorio, Dipartimento Medicina Clinica e Sperimentale, Universita' Di Sassari, Sassari, Italy ; ² Servei De Pneumologia, Hospital Universitari Vall D'Hebron, Barcelona, Spain ; ³ Institut D'Investigacions Biomèdiques August Pi I Sunyer (IDIBAPS), Hospital Clínic, Barcelona, Spain ; ⁴ Servicio De Neumología, Complejo Hospitalario Universitario, Vigo, Spain ; ⁵ Servicio De Neumología, Hospital Universitario, Burgos, Spain and ⁶ Servicio De Neumología, Complejo Hospitalario, Navarra, Spain.

Body: Alpha-1-antitrypsin deficiency (AATD) is an autosomic codominant inherited disorder associated to early onset development of emphysema, that results in significant impairment of health-related quality of life (HRQoL). The aim of this study was to assess the quality of life of patients with emphysema-related AATD compared with a group of patients with smoking COPD. Methods: Observational, cross-sectional study in patients with a severe alpha1-antitrypsin deficiency (phenotype PI*ZZ) and a control group of COPD. All patients completed the mMRC dyspnea scale, the COPD severity score (COPDSS) and the quality of life questionnaires EQ-5D, LCOPD and CAT. Results: A total of 35 PI*ZZ patients were included (mean age 56.5 yrs (SD=10.6)), 57.1% male and mean FEV1(%)= 48.7% (SD=17.9%), and 61 COPD (70.3 yrs (9.2)), 80.3% men and FEV1(%)= 47% (16.4%). Scores of the severity and HRQoL questionnaires were similar, with a tendency towards worse scores in PI*ZZ for the EQ-5D (VAS) 64.8 (20.2) vs 71.6 (17.1); p=0.08. Correlations of HRQoL scores and FEV1(%) predicted were significant in both groups for COPDSS and LCOPD, but not for CAT and EQ-5D index. Correlations were stronger for AATD patients: r= -0.570, p<0.01 for COPDSS and FEV1(%), compared with r= -0.260, p<0.05 for COPD patients. Regarding LCOPD and FEV1(%): r= -0.502, p<0.001 for AATD and r= -0.304, p<0.05 for COPD. Conclusions: The relationship between severity of lung disease and HRQoL, both generic and specific, is stronger in emphysema associated with AATD compared with smokers COPD. The different impact of the disease may be related to the differential characteristics of AATD patients (younger age, lower smoking consumption and less comorbidities).