European Respiratory Society
Annual Congress 2013

Abstract Number: 3578

Publication Number: P464

Abstract Group: 1.5. Diffuse Parenchymal Lung Disease

Keyword 1: Interstitial lung disease **Keyword 2:** Imaging **Keyword 3:** Chronic disease

Title: Familial pulmonary fibrosis: Clinical-functional and radiological features

Dr. David 21784 Bennett david.btt@gmail.com MD [1], Dr. Maria Antonietta 21785 Mazzei mariaantonietta.mazzei@unisi.it MD [2], Dr. Fabrizio 21786 Mezzasalma fabrmezza@virgilio.it MD [1], Dr. Rosa Metella 21787 Refini rosa.refini@unisi.it MD [1], Dr. Elena 21788 Bargagli bargagli2@gmail.com MD [1], Prof. Dr Luca 21789 Volterrani luca.volterrani@unisi.it MD [2] and Prof. Dr Paola 21797 Rottoli rottoli@unisi.it MD [1]. [1] Respiratory Diseases Unit, Dpt. of Medical, Surgical Sciences and Neurosciences, University of Siena, Siena, Italy, 53100 and [2] Radiodiagnostic Unit, Dpt. of Medical, Surgical Sciences and Neurosciences, University of Siena, Siena, Italy, 53100 .

Body: Familial pulmonary fibrosis (FPF) is defined as an idiopathic interstitial pneumonia in two or more consanguineous. Starting from our database, we performed a retrospective analysis of clinical, functional and HRCT features of 35 patients (15 males, age at the diagnosis 58.9±9.2 years), belonging to 23 different families with FPF. Prevalent symptoms at onset were dry cough and dyspnoea, but 10% of the patients were asymptomatic and referred to us for ILD assessment because of their familiar history of pulmonary fibrosis. Two main HRCT patterns have been documented: possible UIP (20/35 patients; 57%) and fibrotic-NSIP (14/35 patients; 40%), only one patient showed a typical UIP pattern. In CT analysis hiatal hernia and mediastinal lipomatosis were statistically more frequent in patients with possible UIP pattern than in fibrotic NSIP ($p<0.001$). Pulmonary function test parameters revealed FEV1 78.8±19.7%; FVC 74.04±19.38%; Tiff 83.5±5.3% and DLCO 50.5±22.8%. Dividing the population according to HRCT findings no difference was found in PFTs parameters and DLCO% ($p>0.05$) between probable UIP and fibrotic NSIP. Following the patients routinely with PFTs every 3 months we observed that no significant worsening in FVC%, FEV1% and DLCO% was evident after 1-year follow-up. Our study shows that functional deterioration is less evident in FPF than expected for sporadic IPF, suggesting that the disease progression is slower. FPF does not present with a single HRCT pattern, being possible UIP and fibrotic NSIP the prevalent ones. Interestingly, hiatal hernia and mediastinal lipomatosis were more frequently observed in the possible UIP group.