

## Challenges and novelties in rare interstitial lung diseases

### Rare ILD/DPLD

#### OA-66852 - Phenotypic characterization of interstitial lung disease associated with mutations in SFTPC and ABCA3 in adults

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**Introduction:** Mutations in surfactant genes SFTPC or ABCA3 are responsible for interstitial lung disease (ILD) in children and adults. Despite numerous paediatric data, few studies described these entities in adults.

**Methods:** We conducted a multicenter retrospective study of adult patients with ILD associated with mutations in SFTPC or ABCA3 in the French rare pulmonary diseases network OrphaLung.

**Results:** We included 36 patients, 22 in the SFTPC group and 14 in the ABCA3 group. Clinical characteristics were similar between the two groups. Baseline pulmonary function tests reported a median FVC of 59% ([52 – 72] %), a



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median FEV1 of 63% ([48,3 – 73,3] %), a median TLC of 65 % ([59 – 76] %) and a median DLco of 44 % ([35,4 – 50] %). A pattern of unclassifiable fibrosing ILD was the most frequent on chest computed tomography, found in 85% of patients. Nonspecific interstitial pneumonia and usual interstitial pneumonia were the most common histological patterns in the ABCA3 group and in the SFTPC group, respectively. Median survival before death or lung transplantation was 10 years in the SFTPC group (4 deaths and 8 patients with lung transplantation) and was not reached at the end of follow-up in the ABCA3 group (1 death and 2 patients with lung transplantation).

**Conclusion:** Patients with ILD and mutations in SFTPC or ABCA3 present a distinct phenotype of unclassifiable fibrosing ILD with cysts and ground glass opacities, and their prognosis is often severe. While our results highlight the phenotypical heterogeneity of these patients, they also expand our knowledge of these rare causes of ILD in adults.

**Keywords:** Orphan diseases, Mutations