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Novel rare genetic variants in idiopathic pulmonary fibrosis

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The study by Nathan and co-workers on rare surfactant protein A genetic variants in patients with familial IPF illustrates the power of rare disease research consortia, and highlights the challenges in developing individualised approaches for IPF patients https://bit.ly/35uC5lw

Cite this article as: Kristof AS. Novel rare genetic variants in idiopathic pulmonary fibrosis. *Eur Respir J* 2020; 56: 2003252 [https://doi.org/10.1183/13993003.03252-2020].

Idiopathic pulmonary fibrosis (IPF) is a rare disease of epithelial injury leading to persistent fibrosis, remodelling of the lung parenchyma, and chronic respiratory failure [1]. IPF can be differentiated from other forms of interstitial fibrosis by the presence of radiological and histopathological features of usual interstitial pneumonia (UIP), and the absence of known initiators (*e.g.* occupational dusts, autoimmunity; reviewed in [2]). Despite attempts to define IPF as a uniform disease entity based on clinical and radiological criteria [3], clinicians continue to struggle with significant variability in radiological appearance, disease progression and response to interventions, which can lead to delays in diagnosis or uncertainty in patient management. This "heterogeneity" arises from genetic and environmental influences which, despite major advances in the field, have thus far been incompletely characterised. As is the case for other rare diseases, policy makers and patients have advocated for the accelerated development of individualised approaches to patient management [4, 5]. Promising advances in the field include the ability to comprehensively characterise molecular and genetic features of disease using state-of-the art technologies, and orphan disease policies that facilitate the development of diagnostic and therapeutic modalities in the public and private sectors [6].