European Respiratory Society statement on familial pulmonary fibrosis

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Genetic predisposition to interstitial lung diseases (ILDs) is well known. The statement aims to assist health professionals with the care of monogenic pulmonary fibrosis and familial ILD patients and their relatives. https://bit.ly/3h9hvQ6

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Abstract

Genetic predisposition to pulmonary fibrosis has been confirmed by the discovery of several gene mutations that cause pulmonary fibrosis. Although genetic sequencing of familial pulmonary fibrosis (FPF) cases is embedded in routine clinical practice in several countries, many centres have yet to incorporate genetic sequencing within interstitial lung disease (ILD) services and proper international consensus has not yet been established. An international and multidisciplinary expert Task Force (pulmonologists, geneticists, paediatrician, pathologist, genetic counsellor, patient representative and librarian) reviewed the literature between 1945 and 2022, and reached consensus for all of the following questions: 1) Which patients may benefit from genetic sequencing and clinical counselling? 2) What is known of the natural history of FPF? 3) Which genes are usually tested? 4) What is the evidence for telomere length measurement? 5) What is the role of common genetic variants (polymorphisms) in the diagnostic workup? 6) What are the optimal treatment options for FPF? 7) Which family members are eligible for genetic sequencing? 8) Which clinical screening and follow-up parameters may be considered in family members? Through a robust review of the literature, the Task Force offers a statement on genetic sequencing, clinical management and screening of patients with FPF and their relatives. This proposal may serve as a basis for a prospective evaluation and future international recommendations.