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| **The utility of genetic testing in adults with pulmonary fibrosis: A retrospective cohort study.** |
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| **Introduction/Aim:** There is increasing recognition of the role that genetic factors play in the aetiology of pulmonary fibrosis (PF). However, confirmatory testing is rarely performed in clinical practice, which may reflect the paucity of evidence-based guidelines in this setting. The aim of this study was to assess the clinical utility of genetic testing in adults with PF within our local cohort. **Methods:** A retrospective cohort study was performed, including all adults with PF who completed genetic testing at a single tertiary centre between 29/6/17 and 27/6/23. **Results:** Forty-one adults with PF completed genetic testing between 29/6/17 and 27/6/23. Twenty-two patients (53.7%) had at least one mutation identified on genetic testing. Of these, nine were considered pathogenic, thirteen were classified as variants of uncertain significance. Four patients had two or more mutations detected. There were nineteen mutations affecting telomere-related genes (TERT, PARN, RTEL1, TERC) six mutations affecting genes involved in surfactant homeostasis (ABCA3) and three mutations in genes associated with PF though alternative mechanisms (SMPD1, ASAH1, HPS4). There was no family history present in eight of the patients with genetic mutations present. Peripheral blood mononuclear cell flow-FISH telomere length (TL) was above the 10th percentile in three patients with mutations in telomere-related genes.**Conclusion:** Amongst our cohort of adult patients with PF, genetic mutations were frequently identified and were not uniquely seen amongst patients with a positive family history. Variants of uncertain significance were commonly identified, which require further assessment to determine pathogenicity. **Grant Support:** Nil.**Key words:** familial pulmonary fibrosis, telomere, surfactant, genetic. |