WTDTP Projects for September 2023

Project Reference: T1/53

Project Title: **The role of rare genetic variants in progressive pulmonary fibrosis**

Theme(s): Theme 1: Genomics for drug development & pharmacogenetics

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Project Summary: Idiopathic pulmonary fibrosis (IPF) is a devastating disease where the lungs become progressively scarred. There is currently no cure for IPF and around 6,000 people are diagnosed with IPF each year. Around half of these individuals will die within three years, however, not everybody experiences the same severity of the disease. Some individuals will experience a rapid decline in health and short survival time, whereas others may live for many years with relatively stable lung health. We have recently identified common genetic changes that are associated with how quickly IPF progresses. We know rare genetic changes can affect how likely an individual is to develop IPF, but we do not know what effect these rare genetic variants have on disease progression.

Through large-scale statistical analyses, this project will investigate whether rare genetic variants and related genes are associated with progressive forms of pulmonary fibrosis. Bioinformatic follow-up analyses will explore how these genes may cause more progressive fibrosis. This will aid our understanding the underlying biological processes involved in progressive pulmonary fibrosis and help the development of desperately needed novel treatments.