

Scientists unlock genetic code of diseased lung cells to find new treatments for IPF

December 8 2016

Researchers cracked the complete genetic code of individual cells in healthy and diseased human lung tissues to find potential new molecular targets for diagnosing and treating the lethal lung disease Idiopathic Pulmonary Fibrosis (IPF).

A team of scientists from Cincinnati Children's Hospital Medical Center, in collaboration with investigators at Cedars-Sinai Medical Center in Los Angeles, publish their findings Dec. 8 in the *Journal of Clinical Investigation Insights (JCI Insights)*.

"This paper identifies a number of novel targets and molecular pathways for IPF, for which there are pharmaceutical approaches," said Jeffrey Whitsett MD, lead investigator and co-director of the Perinatal Institute at Cincinnati Children's. "Airway cells can be obtained by brushing the airway or biopsy, and marker genes can be tested to make a diagnosis or monitor treatment."

IPF is a common and lethal interstitial [lung](#) disease in adults, which means it inflames, scars and reconfigures lung tissues. This causes loss of the air sacs, called alveoli, where oxygen and carbon dioxide are normally exchanged. Similar losses of [lung function](#) can occur earlier in life, especially in children with diseases caused by mutations in genes critical for surfactant and maintenance of the lung saccules.

Biological processes controlling the formation and function of the lung's alveolar region require precisely orchestrated interactions between

diverse epithelial, stromal and immune cells, according to study authors. Despite many years of extensive laboratory studies of whole tissue samples - trying to identify genetic, cellular and molecular processes that fuel [lung ailments](#) like IPF - the precise biology has remained elusive.

To overcome this, Whitsett and colleagues - including first author and bioinformatician Yan Xu, PhD of Cincinnati Children's - conducted what they believe to be the first-ever single-cell RNA sequence analysis of normal and diseased human lung tissues (all donated with prior informed consent). This provided the authors with a detailed genetic blueprint of all the different epithelial cell types involved in IPF progression and a window to identify aberrant biological processes driving inflammation and fibrosis.

Analysis of normal lung epithelial cells found gene patterns linked to fully formed alveolar type 2 [lung cells](#) (AT2 cells), which are important for the production of surfactant, a substance containing a complex of proteins critical to breathing.

Analysis of diseased IPF cells found genetic markers for lung cells that were in indeterminate states of formation, the authors report. IPF [cells](#) had lost the normal genetic control systems needed to guide their functions. This study identifies abnormalities in gene expression that can be targeted for therapy of [chronic lung diseases](#) like IPF.

More information: Yan Xu et al, Single-cell RNA sequencing identifies diverse roles of epithelial cells in idiopathic pulmonary fibrosis, *JCI Insight* (2016). [DOI: 10.1172/jci.insight.90558](https://doi.org/10.1172/jci.insight.90558)

Provided by Cincinnati Children's Hospital Medical Center

Citation: Scientists unlock genetic code of diseased lung cells to find new treatments for IPF (2016, December 8) retrieved 9 April 2024 from <https://medicalxpress.com/news/2016-12-scientists-genetic-code-diseased-lung.html>

This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.