MedicalXpress

Genetic risk profile predicts survival for people with severe lung disease

September 20 2017

An international Yale-led research team has shown that a risk profile based on 52 genes accurately predicts survival for patients with a severe lung disease. If confirmed in further studies, the finding could transform the way patients are treated for the condition, which is on the rise in older adults.

The study published in *The Lancet Respiratory Medicine*.

Idiopathic pulmonary fibrosis (IPF) is a form of lung disease that causes scarring and impairs breathing. Without a transplant, IPF patients can expect to live between three and five years. In a prior study, lead authors Jose Herazo-Maya M.D., Naftali Kaminski, M.D., and colleagues identified 52 genes that correlated with survival. However, they only validated four of those genes and on a limited number of patients.

In the new study, the Yale team collaborated with researchers in the United Kingdom, Germany, and the United States to determine if the entire 52-gene risk profile could be validated in multiple patient groups. They also assessed whether the risk profile would enhance tools currently used by clinicians, and whether the risk profile would change as the disease progresses.

The research team collected samples from more than 400 patients recruited from multiple academic centers over a decade. The researchers developed a system to score genes with high or low expression, and classified patients as being at either high or low risk for IPF mortality.

To determine the predictive value of the risk profile, they also analyzed patient health data and response to drug therapy over time.

The researchers found that the 52-gene risk profile did accurately predict individual patient survival, in all six cohorts, said Herazo-Maya. "IPF is known to have a variable and unpredictable course and therefore we were so impressed that the results were validated in all six cohorts," he noted.

The gene risk profile increased the accuracy of current clinical tools. The study also found that the risk profile worsens as the disease progresses. In a small subset of patients treated with recently FDA-approved medications, the risk profile improved.

While further study is needed, the findings have important implications, said the researchers. The 52-gene risk profile may be useful to make disease management decisions, such as prioritizing some patients for lung transplantation, said Kaminski. "If additional studies confirm the treatment effect we observed, shifts in the 52-gene risk profile could eventually be used to indicate response to therapy, and allow more efficient drug trial designs and disease management plans," he explained.

Provided by Yale University