



Cystic Fibrosis Research News

Citation:

Corvol H, Blackman SM, Boëlle PY, et al. Genome-wide association meta-analysis identifies five modifier loci of lung disease severity in cystic fibrosis. *Nat Commun.* 2015 Sep 29; 6:8382

What was your research question?

What are non-cystic fibrosis transmembrane conductance regulator (CFTR) genes that affect lung disease severity in cystic fibrosis?

Why is this important?

CF is caused by a mutation in the CFTR gene, and new therapies targeting the CFTR protein can have a significant impact on the health of individuals with CF. However, lung disease can also be influenced by factors outside of the CFTR gene and may require more individualized treatment. Identification of other genes that can affect lung disease may provide additional treatment targets that differ from person to person.

What did you do?

This study investigated over 8 million genetic variants (DNA differences) in over 6,365 individuals with CF. We calculated a measure of lung function called the Kulich normal residual, mortality adjusted (KNorMA) for each person. This KNoRMA number allows for comparison of disease progression and rates of death regardless of age and gender. Genetic variants were analysed to identify regions of DNA that modify CF lung disease.

What did you find?

Five regions of genetic material (DNA) were identified that associate with differences in the KNoRMA (measure of lung function). These regions contain genes that could be related to the development or severity of lung disease.

What does this mean and reasons for caution?

Identification of factors that modify CF lung disease could help in providing new targets for CF treatment. This information could also aid in our understanding of how the disease develops and progresses.



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What's next?

Further investigation and a functional study of genes identified is necessary to help us better understand how to use these findings to target specific treatments.