

Cystic Fibrosis Research News

Citation:

Aksit MA, et al. Genetic Modifiers of Cystic Fibrosis-Related Diabetes Have Extensive Overlap with Type 2 Diabetes and Related Traits. J Clin Endocrinol Metab. 2020

What was your research question? (50 words maximum)

We wanted to identify genetic modifiers of CFRD (cystic fibrosis related diabetes) and determine the genetic overlap with other types of diabetes.

Why is this important? (100 words maximum)

Many patients develop CF related diabetes, which is distinct from other types of diabetes. It is also associated with worse outcomes of individuals living with CF. The risk for CFRD is determined by CFTR (cystic fibrosis transmembrane regulator) genotype as well as other variants. All are important to identify for prognostic and therapeutic purposes.

What did you do? (100 words maximum)

A genome-wide association study was performed for CFRD onset in over 5700 individuals with CF.

What did you find? (100 words maximum)

Three loci reached genome-wide significance: PTMA (novel) and TCF7L2 and SLC26A9 (previously identified). CFRD associates strongly with polygenic risk scores for type 2 diabetes and less so for type 1 diabetes.

What does this mean and reasons for caution? (100 words maximum)

CFRD and type 2 diabetes are more closely related than previously realized; however, two CFRD loci are unrelated to type 2 diabetes and may affect more aspects of CF than the development of CF-related diabetes.

What's next? (50 words maximum)

Replication of these findings and further investigation into the CFRD-related loci is needed.