

Cystic Fibrosis Research News

Title:

Cystic Fibrosis Transmembrane Conductance Regulator Gene Variants are Associated with Autoimmune Pancreatitis and Slow Response to Steroid Treatment

Authors:

Ming-Chu Chang, MD, PhD¹, I-Shiow Jan, MD², Po-Chin Liang, MD², Yung-Ming Jeng, MD, PhD³, Ching-Yao Yang, MD, PhD⁴, Yu-Wen Tien, MD, PhD⁴, Jau-Min Wong, MD, PhD¹, Yu-Ting Chang, MD, MS, PhD¹

Affiliations:

¹Department of Internal Medicine, National Taiwan University Hospital, College of Medicine, National Taiwan University, Taipei, Taiwan; No.7 Chung Shan South Road, Taipei, Taiwan

²Department of Laboratory Medicine, National Taiwan University Hospital, College of Medicine, National Taiwan University, Taipei, Taiwan; No.7 Chung Shan South Road, Taipei, Taiwan

³Department of Pathology, National Taiwan University Hospital, College of Medicine, National Taiwan University, Taipei, Taiwan; No.7 Chung Shan South Road, Taipei, Taiwan

⁴Department of Surgery, National Taiwan University Hospital, College of Medicine, National Taiwan University, Taipei, Taiwan; No.7 Chung Shan South Road, Taipei, Taiwan

What was your research question?

To consider whether different variants of the cystic fibrosis transmembrane conductance regulator (CFTR) gene are linked to the condition of autoimmune pancreatitis.

Why is this important?

Genetic mutations in the CFTR gene mean that it doesn't function properly and this is linked to chronic pancreatitis (inflammation of the pancreas), including chronic pancreatitis of unknown causes. Autoimmune pancreatitis is a distinct form of chronic pancreatitis, where

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the body's own immune cells attack the pancreas and stop it functioning. Until now, there have not been comprehensive investigations looking at whether different variants of the CFTR genes are linked to autoimmune pancreatitis and whether the presence of these different versions affects patients' clinical course.

What did you do?

Variants of the CFTR gene were carefully examined using next-generation sequencing in samples taken from 89 people with autoimmune pancreatitis. We compared the clinical features of those people with autoimmune pancreatitis who also had mutations of the CFTR gene and those who didn't. These features included imaging, tissue samples, blood samples, how fast they respond to treatment with steroids and other issues with organs other than the pancreas e.g. liver.

What did you find?

A total of 28.1% (25/89) of the people with autoimmune pancreatitis carried 26 CFTR mutations. These mutations are predictors of a slower and a reduced response to steroid treatment in people with autoimmune pancreatitis. In addition, next-generation sequencing is a powerful approach for rapidly screening different CFTR mutations at the same time.

What does this mean and reasons for caution?

This study showed that other treatments for suppressing or moderating the immune system such as Imuran or anti-CD20 should be considered as a first-line treatment instead of steroids in people with autoimmune pancreatitis who have with CFTR mutations. Further multinational studies (run across a number of different centres so as to increase the amount of information available) should be performed to confirm our findings.

What's next?

Larger genetic studies of other ethnic populations and studies looking at CFTR mutations and how they work at a basic cell level will offer additional information regarding how CFTR mutations affect autoimmune pancreatitis.



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cfresearchnews@gmail.com