



Cystic Fibrosis Research News

Title:

Assessment of Epithelial Sodium Channel Variants in Nonwhite Cystic Fibrosis Patients with Non-Diagnostic CFTR Genotypes

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What was your research question?

We explored whether genetic changes (mutations) of the Epithelial Sodium Channel (ENaC) genes are a cause of cystic fibrosis (CF) in nonwhite people with CF.

Why is this important?

Cystic fibrosis (CF) is conventionally considered to be caused by mutation of a single gene, the *CF Transmembrane Conductance Regulator* (CFTR) gene. The vast majority of people with CF have recognised mutations of their CFTR genes, but in a small but significant number of people there is no recognised mutation. This is particularly noticeable in nonwhite people with CF and made us wonder whether their condition may relate to the mutation of a different gene.

It is important to know what mutations people with CF have as this may help them with pregnancy choices and identify whether they may benefit from new therapies like Ivacaftor which target specific mutations.

What did you do?

We explored the Epithelial Sodium Channel (ENaC) genes which make a protein that is involved in salt transport in a similar way to the CFTR gene. We looked carefully for well-recognised mutations and also other less well-recognised mutations of this gene in nonwhite people with CF.

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What did you find?

We did not find any evidence to suggest that a mutation of the ENaC genes causes CF-like disease in this population.

What does this mean and reasons for caution?

Although this is a small study, we found that a mutation of the ENaC genes is not a common cause of CF-like disease in nonwhite people with CF in our study group. It may be that they have, as yet, unrecognised mutations of their CFTR genes or they may have mutations of other genes.

What's next?

Future studies should examine these genes more carefully, explore other genes and involve larger numbers of people with CF to better identify these mutations.

Original manuscript citation in PubMed

http://ac.els-cdn.com/S1569199315001010/1-s2.0-S1569199315001010-main.pdf?_tid=a324a2f6-1971-11e5-b5f8-00000aacb35e&acdnat=1435041336_49234144b79469164e4c047c5e9a2f66