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Cystic Fibrosis Research News

Title:
Non-allergic asthma as a CFTR-related disorder

Authors:
Angela Schulz[1,2], Burkhard Tümmler[1,2]

Affiliations:
[1]Clinic for Paediatric Pneumology, Allergology and Neonatology, Hannover Medical School, Hannover, Germany
[2]Biomedical Research in Endstage and Obstructive Lung Disease (BREATH), German Center for Lung Research, Hannover, Germany

What was your research question?
Cystic fibrosis (CF) is diagnosed if two disease-causing mutations in the *Cystic Fibrosis Transmembrane Conductance Regulator* (*CFTR*) gene associated with elevated chloride concentrations in the sweat test are found. However, some individuals may show some CF-like symptoms, but with no CFTR mutation identified and with a sweat chloride value in a grey zone between normal and a diagnosis of CF.

Why is this important?
If a doctor suspects that the patient's CFTR does not work properly, but CFTR genetics and sweat test do not give a clear answer, she/he will send the patient to specialists who tests CFTR function in the nose by nasal transepithelial difference (NPD) measurements or in rectal biopsies by intestinal current measurements (ICM).

What did you do?
We have undertaken NPD and ICM measurements according to the protocols of the Diagnostic Working Group of the European CF Society on 102 people with CF-like symptoms, but inconclusive genetics and sweat test results.

What did you find?
Seven of the 102 subjects had been diagnosed previously to suffer from 'non-allergic asthma'. These seven people showed a sweat test in the grey zone, normal CFTR function in the ICM and a reduced CFTR function within the CF range in the NPD. Of the organs tested, CFTR function was only impaired in the airways.

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What does this mean and reasons for caution?

Defects in the CFTR gene do not only cause CF, but can also be involved in acute and chronic diseases of the airways or pancreas and may affect the ability of a man to father children. These latter diseases are classified as CFTR-related disorders. Our study suggests that non-allergic asthma is included in the list of CFTR-related disorders.

What's next?

We studied a highly selected group of people sent to us with the suspected diagnosis of a CFTR-associated disease. Hence next the frequency of this yet undescribed CFTR-related disorder should be investigated among unselected people with non-allergic asthma. Depending on the outcome one will decide of whether or not NPD should be included into the diagnostic routine of non-allergic asthma.

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