Cystic Fibrosis Research News

Title:

Year to year change in FEV1 in patients with cystic fibrosis and different mutation classes.

Authors:

K De Boeck[1], A Zolin[2]

Affiliations:

1.Department of Pediatrics, University of Leuven, Belgium
2.Department of Clinical Sciences and Community Health, University of Milan, Milan, Italy

What was your research question?

Errors (mutations) in the cystic fibrosis (CF) gene can be grouped in 'classes' according to how the error disturbs the production or function of the CFTR (cystic fibrosis transmembrane conductance regulator) protein. There are six classes which describe the errors in the CFTR gene: no production of full length protein (class I); protein brake down (class II); defective protein channel opening (class III); insufficient passage of salts (class IV); insufficient protein production (class V) and instability of the protein in the cell membrane (class VI). We wanted to know whether there is a difference in speed of loss of lung function between people with different classes of mutations.

Why is this important?

That information is important, to understand the 'benefit' of the new very costly mutation class specific medications.

What did you do?

We calculated the current loss of lung function, measured by year to year change in forced expired volume in one second (FEV1) in 11,417 people with CF included in the European Cystic Fibrosis Society Patient Registry.

What did you find?

Whereas individuals with at least one mutation of class IV or V have on average a lower year to year change, we did not find a difference between those with a stop codon mutation, homozygous for F508del or at least one class III mutation.

Cystic Fibrosis Research News

What does this mean and reasons for caution?

This means there is no difference in loss of lung function between the latter groups. It may be important to measure over a longer time to be certain there is no difference at all between these groups.

What's next?

We plan to explore the registry data further and over a longer time period, to be certain about the conclusion.

Original manuscript citation in PubMed

https://www.ncbi.nlm.nih.gov/pubmed/?term=Year+to+year+change+in+FEV1+in+patients+with+cystic+fibrosis+and+different+mutation+classes.