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Cystic Fibrosis Research News

Title:

Clinical and Functional Efficacy of Elexacaftor/Tezacaftor/Ivacaftor in People with Cystic Fibrosis Carrying the N1303K Mutation

Lay Title:

Elexacaftor/Tezacaftor/Ivacaftor improves CFTR activity and pulmonary function in Cystic Fibrosis Patients carrying the N1303K Mutation

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What was your research question?

Does treatment with elexacaftor/tezacaftor/ivacaftor (ETI) improve lung function and other clinical parameters in individuals with cystic fibrosis carrying the N1303K mutation?

Why is this important?

The N1303K mutation is the third most common mutation in people with cystic fibrosis (pwCF) and is associated with classic CF symptoms including pancreatic insufficiency and chronic lung damage. Unfortunately, this mutation was not considered responsive to a particular ETI treatment, so patients with this mutation were unable to benefit from it. However, our study has shown strong evidence that individuals with the N1303K mutation can actually experience significant improvements when treated with ETI. This means that there is potential for positive health outcomes and more treatment options for this specific group of cystic fibrosis patients.

What did you do?

We collected clinical data involving eight pwCF with at least one copy of the N1303K mutation. The study aimed to assess the effects of ETI treatment on these individuals. The data was gathered both before and after an eight-week course of ETI. Additionally, we conducted tests using cells from their intestines to see how they reacted to the medication.

What did you find?

After starting ETI treatment, all eight pwCF showed significant improvements in various aspects of their health. On average, their lung function increased by 18.4%, which translates to a 26.5% improvement from the initial measurements. Their body mass index also increased by an average of 0.79 Kg/m2. The lung clearance index decreased by 3.6 points, indicating a 22.2% improvement. However, there were no notable changes in sweat chloride levels. Four patients had their nasal potential difference return to normal, and tests using specialized cells showed improved activity of CFTR channels.

What does this mean and reasons for caution?

Our study indicates that ETI treatment may provide benefits to pwCF carrying the N1303K mutation. We observed statistically significant improvements in several clinical parameters, suggesting the effectiveness of the treatment. However, it is important to note that our study had a limited sample size, which means we should be cautious in extending these findings to a larger population.

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What's next?

Our study shows that ETI treatment could be beneficial for pwCF who have the N1303K mutation. We saw significant improvements in their health. Further research is needed to evaluate the long-term effects of the ETI treatment, such as exacerbation rates, sputum bacterial growth, and other relevant clinical outcomes.

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