Cystic Fibrosis Research News

Title:
Every CFTR variant counts – Target-capture based Next-Generation-Sequencing for Molecular Diagnosis in the German CF Registry

Lay Title:
Unlocking the Secrets of Cystic Fibrosis: A Genetic Study of people with CF in Germany

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What was your research question?

We aimed to better understand cystic fibrosis (CF) by improving the methods we are using to identify the gene mutations that cause CF (within a gene called *CFTR*). Knowing the exact mutations can help doctors pick the best treatments.

Why is this important?

CF is a serious genetic disorder, and treatment plans can differ based on the exact gene mutations a person with CF has. Understanding these genetic details can help doctors make the best treatment choices and consequently improve quality of life for people with CF. Currently, we focus our analysis on only a portion of the gene that causes CF, but mutations can occur within the whole gene. So in order to identify the gene mutations for all people with CF it is important to also explore the regions that are not studied in traditional genetic analyses.

What did you do?

We used advanced DNA testing methods, called Next-Generation Sequencing (NGS), to look at the entire CFTR gene. We studied 655 people with CF who gave consent for the German CF Registry. We identified not only common mutations but also some that had never been seen before. We also corrected errors in the Registry data to ensure that the information is accurate and up-to-date.

What did you find?

Our in-depth exploration of the CFTR gene of hundreds of people with CF revealed several important findings. First, many people had mutations that are not typically tested for, underlining the need for comprehensive genetic testing. Second, we found 43 new mutations never described before. This could potentially change how we understand and treat people

with CF. Third, a significant number of mutations were uncertain, requiring further studies to understand their impact on a person's health.

What does this mean and reasons for caution?

Our study provides a more complete picture of CF gene mutations in Germany. This can help in personalized treatment plans, inform family members who might be at risk, and help in developing new therapies. However, our methodology has its limitations and not all mutations we found are fully understood yet.

What's next?

Next, we recommend a two-step genetic testing strategy: an initial basic test followed by a comprehensive one if needed. This could speed up finding the right treatments for people with CF, especially those with rare mutations.

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