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Cystic Fibrosis Research News

Title:

Theranostics by testing CFTR modulators in patient-derived materials: the current status and a proposal for subjects with rare *CFTR* mutations

Authors:

Margarida Amaral[1], Kris De Boeck[2]
On behalf of the ECFS Strategic Planning Task Force on 'Speeding up access to new drugs for CF': Margarida Amaral[1], Kris De Boeck[2], Jane C Davies[3], Pavel Drevinek[4], Stuart Elborn[5], Eitan Kerem[6], Tim Lee[7]

Affiliations:

1) University of Lisboa, Portugal
2) University of Leuven, Leuven, Belgium
3) National Heart and Lung Institute, Imperial College, London, United Kingdom
4) Motol University Hospital, Prague, Czech Republic
5) Queen's University, Belfast, United Kingdom
6) Hadassah Medical Center, Jerusalem, Israel
7) Leeds Children's Hospital, Leeds, United Kingdom

What was your research question?

Can we speed up drug development for people with cystic fibrosis (PWCF) who have a rare or very rare mutation.

Why is this important?

Development of drugs that treat the basic defect in CF is at present very much focused on mutation F508del or on mutations that are relatively common. But we must of course make a plan to help every person with CF, also those people who have a very rare mutation.

What did you do?

We convened a workshop with relevant stakeholders (researchers, doctors, patient organizations, pharma companies, regulators) and discussed the different methods that are available to test the benefit of drugs directly on the tissues of PWCF, with all their pro's and con's.

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What did you find?

The drugs that treat the basic defect in PWCF with common mutations, can be tested directly on the patient's own tissue if it concerns a person with CF who has a rare or even unique mutation. Starting with material obtained from scrapings taken from the patient's nose or small painless biopsies taken from the patient's rectum is the most practical way forward. There is already quite a bit of evidence that these tests help to identify the right treatment for the right person.

What does this mean and reasons for caution?

This brings promise for PWCF and rare CFTR mutations. Since these people are spread to all corners of the world, organizing this testing on many patients will require a big effort from the entire community (patients, doctors and researchers) . The other hurdle is the high cost of the current medications. But of course knowing who will be a good candidate for a specific treatment is an important first step.

What's next?

Testing this plan on a larger scale such as is done in the ambitious European HIT CF program (https://www.hitcf.org) where patients with very rare mutations will have first testing on their tissue and if a benefit is seen they are invited to participate in a clinical trial.

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cfresearchnews@gmail.com