Cystic Fibrosis Research News

Title:
Newborn screening alone insufficient to improve pulmonary outcomes for cystic fibrosis

Authors:
Christina B. Barreda\textsuperscript{a}, Philip M. Farrell\textsuperscript{a}, Anita Laxova\textsuperscript{a}, Jens C. Eickhoff\textsuperscript{b}, Andrew T. Braun\textsuperscript{c}, Ryan J. Coller\textsuperscript{a}, Michael J. Rock\textsuperscript{a}

Affiliations:
\textsuperscript{a} Department of Pediatrics, University of Wisconsin-Madison School of Medicine and Public Health, 600 Highland Ave, Madison, WI 53792, USA
\textsuperscript{b} Department of Biostatistics and Medical Informatics, University of Wisconsin-Madison School of Medicine and Public Health, 600 Highland Ave, Madison, WI 53792, USA
\textsuperscript{c} Department of Medicine, University of Wisconsin-Madison School of Medicine and Public Health, 600 Highland Ave, Madison, WI 53792, USA

What was your research question?
In an era prior to strict infection control and current therapies, did patients with cystic fibrosis (CF) without meconium ileus who were identified by newborn screening (screened group) have better long-term pulmonary and mortality outcomes compared with those who presented clinically (control group)?

Why is this important?
Before current therapies and infection control measures were in place, it was not known if early diagnosis of CF by newborn screening would be beneficial. A previous prospective randomized control trial followed infants with CF born in the late 20\textsuperscript{th} century in the state of Wisconsin (USA) and found that those identified by newborn screening (screened group) had better nutritional outcomes compared to those who were diagnosed later after presenting with symptoms (control group). However, there was no clear difference in lung function up to age 21 years. Now that these patients are older, this study aimed to compare the longer term pulmonary and mortality outcomes of the two groups.

What did you do?
Since nearly all of the patients in the original study have clinical data recorded in the Cystic Fibrosis Foundation Patient Registry (CFFPR), we obtained information on their clinical characteristics, lung function, and mortality. We then compared the course of lung disease by evaluating pulmonary function data over time and mortality outcomes of the two groups.
Cystic Fibrosis Research News

Taking advantage of its proven reliability, we focused on percent predicted forced expiratory volume in 1 second (ppFEV1) and mortality. Appropriate statistical modelling was used to compare the ppFEV1 decline of subjects.

What did you find?
We found that the patients identified with CF by newborn screening had a greater decline in ppFEV1 up to age 26 years old compared to the patients who were diagnosed later after presenting with symptoms. Early infection with Pseudomonas aeruginosa prior to 2 years of age due to exposures in a mixed, integrated clinic caring for older, infected patients simultaneously appeared to contribute to the worse results in the screened group. There was no difference in mortality between the two groups, but at 25 years of age, 89% of screened group enrollees and 85% of the control patients were still alive, thus limiting our ability to compare this outcome measure.

What does this mean and reasons for caution?
Newborn screening by itself is not sufficient to improve pulmonary outcomes for people with cystic fibrosis. It is important to note that the patients in this study grew up in the late 20th century, prior to the advent of current infection control standards and therapies for CF. Very early acquisition of Pseudomonas aeruginosa appeared to be part of the reason that the screened group had worse pulmonary outcomes, so these results might not be seen in patients who entered the health care setting after strict infection control measures were put in place.

What’s next?
Researchers should continue to monitor the long-term outcomes of patients identified by newborn screening to adjust management (such as improving infection control) when unforeseen consequences arise.

Original manuscript citation in PubMed