Journal of Cystic Fibrosis
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Cystic Fibrosis Research News

Citation:

Ren CL, Fink AK, Petren K, Borowitz DS, McColley SA, Sanders DB, Rosenfeld M, Marshall BC. Outcomes of Infants with Indeterminate Diagnosis Detected by Cystic Fibrosis Newborn Screening. Pediatrics. 2015 Jun;135(6):e1386-92. PMID: 25963008.

What was your research question? (50 words maximum)

The goal of this study was to determine the number of cases, clinical features, and short-term outcomes of infants with cystic fibrosis (CF) transmembrane conductance regulator-related metabolic syndrome (CRMS).

Why is this important? (100 words maximum)

There is very little information available on the disease progression and outcomes of CRMS. CRMS describes infants that have a positive newborn screen for CF, but do not show symptoms and show inconclusive diagnostic testing (for example, a borderline sweat test value). Infants without a concrete diagnosis present a treatment challenge to doctors and can create added stress for families. Better understanding of this population can offer more knowledge of how to care for this population.

What did you do? (100 words maximum)

We looked at data from the U.S. CF Foundation Patient Registry (CFFPR) for the years 2010 to 2012. We compared demographics, diagnostic information, population size, health care use, bacteria cultured, and treatment characteristics of infants with CF and infants with CRMS..

What did you find? (100 words maximum)

There were 1,983 infants diagnosed using newborn screening (NBS) reported to the CFFPR between 2010 and 2012. Based off CF Foundation guideline

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definitions, 1,540 infants met criteria for CF and 309 infants met criteria for CRMS. Of the infants with CRMS, 40.8 percent were entered into the registry with a clinical diagnosis of CF. Infants with CRMS tended to have normal nutrition patterns. 11 percent of CRMS infants had a positive Pseudomonas aeruginosa culture within the first year of life. Growth markers did not differ substantially between CF and CRMS infants.

What does this mean and reasons for caution? (100 words maximum)

CRMS is a common outcome following a CF newborn screen. While most infants with CRMS are healthy, a small percentage of them may develop symptoms of CF. These infants should be monitored for the development of clinical CF features. Reasons for caution with these findings include incomplete information for some data categories, CF mutations that are not yet fully characterized, and possible differences in classification of infants with CRMS depending on the doctor or care center.

What's next? (50 words maximum)

These results will be helpful to clinicians who need to counsel and care for infants with CRMS and their families. This was the largest multicenter study to date on infants with CRMS. Further research will be needed to continue understanding diagnosis and treatment for those with CRMS.