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Cystic Fibrosis Research News

Title:

A survey of the prevalence, management and outcome of infants with an inconclusive diagnosis following newborn bloodspot screening for cystic fibrosis (CRMS/CFSPID) in six Italian centres

Lay Title:

Prevalence, management and outcome of Italian infants with cystic fibrosis screening positive inconclusive diagnosis

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What was your research question?

What is the prevalence, management and outcome of infants with an inconclusive diagnosis following newborn bloodspot screening for cystic fibrosis (CRMS/CFSPID) in six Italian centres?

Why is this important?

More information is required on the characteristics and outcomes of infants with cystic fibrosis (CF) who screen positive for CF but the diagnosis is inconclusive symptomatically. This study reports the results from a large group of infants from five regions of Italy using a variety of newborn screening approaches.

What did you do?

We describe the clinical characteristics of 336 children with inconclusive CF diagnosis, born from January 2011 to August 2018. We compared them to 257 infants with CF diagnosed by newborn screening, born in the same period. In each centre, all data were recorded by one expert research assistant (monitor).

What did you find?

The gene profile of the Italian population increases the risk of recognition of infants with inconclusive diagnosis. There is great variability across the six involved centres in the clinical management of infants with an inconclusive CF diagnosis. The majority of the infants were diagnosed as not suffering from CF, except for 5% of patients whose diagnosis was converted to CF. For the majority of those infants diagnosed with CF, it was based on the results of the sweat test becoming positive (more than 59 mmol/L), in the absence of symptoms. This highlights the importance of repeated sweat testing in infants with an inconclusive CF diagnosis, at least every 12 months in the first few years of life.

What does this mean and reasons for caution?

1. The approach to newborn screening can impact significantly on the detection of infants with an inconclusive CF diagnosis.
2. Care for infants with an inconclusive CF diagnosis varies between centres, risking over-testing in many cases.
3. A reassessment of these infants is desirable at three years of age.

What's next?

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More studies are required to guide the management of infants with an inconclusive CF diagnosis, and to know the percentage of them progressing to having a dysfunctional CFTR (malformed protein) related disorder or developing multi organ involvement CF.

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