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Cystic Fibrosis Research News

Title:
When CFSPID becomes CF

Lay Title:
Description of children who changed diagnosis from indeterminate newborn screening to cystic fibrosis

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What was your research question?
We wanted to describe children who initially had the diagnosis of Cystic Fibrosis Screen Positive, Inconclusive Diagnosis (CFSPID) who were later reclassified as having cystic fibrosis (CF).

Why is this important?
Some babies have an abnormal test when they are born that could mean they have a life-limiting disease called CF, and they are given a diagnosis of CFSPID. We currently don't know which of these children will go on to be healthy and which ones might later be diagnosed with CF. We want to be able to help specialists and families have a better understanding of how these screen-positive children might do based on our experience so that children can be diagnosed and treated as early as possible.

What did you do?
We looked at 10 children out of 154 screen-positive referrals over 12 years in our clinic who were initially diagnosed with CFSPID and who later were diagnosed with CF. We looked at

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sweat chloride tests over time to see how the values changed. We studied their DNA glitches, also called variants or genetic mutations, what bacteria they grew, their lung function, and chest images. We described how each of these children did over time and what led CF providers to change their diagnosis to CF.

What did you find?

We found that it is still a challenge to decide what factors contribute to changing a child's diagnosis from CFSPID to CF in children identified by newborn screening. We found that some of the children who were reclassified had an initial sweat test of below 30 mmol/L, which may mean there are some risk associated using with this low cut-off level. These children also had increasing sweat test results of more than 5 mmol/L per year, highlighting the importance of yearly follow-up. In a few children, we used a new test that measures the CF protein function, which helped to define the diagnosis.

What does this mean and reasons for caution?

We explained which factors might be associated with being diagnosed with CF later during childhood. Common factors included specific genetic mutations, growth of Pseudomonas aeruginosa)a bug often found in the lungs of people with CF), and an increase in sweat chloride of more than 5 mml/L per year. These tests do not definitively tell us which children will be diagnosed with CF, and more studies are needed to more clearly define who is at risk. The CF protein function test mentioned above, which is done with cells form the nose (nasal brushing), may be very helpful to make this diagnosis, but more research is needed.

What's next?

As more children with an indeterminate diagnosis continue to be identified by newborn screening, the CF community needs to monitor and report which children are later diagnosed with CF. This will help give better guidance as we study specific trends in larger patient groups.

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