

Cystic Fibrosis Research News

Title: Cystic Fibrosis in Black African Children

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What was your research question?

There is little information on cystic fibrosis (CF) in black African children. We aimed to document CF presentation and outcomes in black African children and hypothesized that it presents similarly to children of European descent in terms of disease onset, severity and long-term outcomes after medical treatment.

Why is this important?

There is a lot of medical information on CF disease in people of European descent worldwide including South Africa. However, very little information on CF disease in black Africans exists. It was previously thought that CF did not exist among black Africans. Failure to diagnose CF in young children leads to avoidable suffering and death. By describing CF in black African children, we hope to increase awareness so that health practitioners and the public will consider CF as a possible cause for malnutrition and chronic chest illness. These similar presentations present commonly in Africa due to other reasons.

What did you do?

We studied all black African children diagnosed with CF in two South African CF centres between January 2000 and March 2018 and compared them to children of similar age, gender and diagnosis date who had two copies of pPhe.508del mutation. We reviewed information of their diagnosis (age, genetics, sweat tests and nutrition), growth, lung function and lung infections over the study period. In children who died, we recorded the cause of death and explored circumstances surrounding these deaths.

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What did you find?

We identified 34 black African diagnosed with CF in the two centres. The average age at diagnosis was 5.5 months and the most common mutation was 3120+1G>A. Compared to the children with the pPhe.508del mutation, black African children were more malnourished and fewer had obstruction of the bowel (meconium ileus) at the time of diagnosis. However, with appropriate care and treatment, growth, lung function and lung infections over time were similar in both groups. Four children died during the study period, 3 black Africans and one in the comparison group. The deaths were soon after diagnosis and related to severe infections from underlying malnutrition.

What does this mean and reasons for caution?

This is the largest report of CF in black Africans and supports the existence of the disease in this cultural group. Unlike children with the pPhe.508del mutation, common in people of European descent, 3120+1G>A was the most common CF mutation among black Africans. Severe malnutrition is the most common mode of presentation in this population. Recognising that CF may be a cause of unexplained malnutrition in infancy is important as children are likely to die from severe infections without appropriate CF treatment. There is a need to continue to raise awareness among clinicians about the existence of cystic fibrosis in Africans.

What's next?

The information learnt from this study has highlighted a need to improve CF diagnosis capacity in Africa and identify other CF mutation in African people so that future diagnostic tests and eventually newborn screening programmes can be developed.

Original manuscript citation in PubMed

<https://www.ncbi.nlm.nih.gov/pubmed/31678015>