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Cystic Fibrosis Research News

Title:

Updated guidance on the care of children with an inconclusive diagnosis after newborn screening

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What was your research question?

To establish optimal care and follow-up for children with an inconclusive diagnosis after a positive newborn screening (NBS) result for cystic fibrosis (CF).

Why is this important?

Identification of infants with an inconclusive diagnosis after a positive NBS result is an increasingly recognised outcome, which leads to uncertainty for both families and healthcare professionals. The approach to the management of these infants is evolving with increased experience and reporting of outcomes. It is important that strategies are based, whenever possible, on evidence of benefit for the child and family. To default these children to a full CF care pathway is not appropriate, but it is equally important that the potential risks that these infants face are recognised and addressed.

What did you do?

A core panel of experts of the European CF Society (ECFS) Neonatal Screening Working Group (NSWG) has investigated all available studies that have cared for and examined children with an inconclusive diagnosis after a positive NBS result for CF and have developed a concept for the care of these children. Subsequently, all core members of the ECFS NSWG and other experts from across the globe have drawn up new guidance for the care of these children in the first 6 years of life.

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What did you find?

The majority of these infants with an unclear diagnosis remain healthy and do not convert to a CF-diagnosis in childhood. However, some children will get a CF diagnosis later on, and monitoring of these infants should facilitate the early recognition of CF. Some infants have a risk to develop a CFTR-related disorder later in life such as congenital bilateral absence of the vas deferens (CBAVD), a well-recognised cause of male infertility, and families need to be provided with clear information about this. The new recommendations include an annual check-up with a more thorough examination at the age of 6 years: then it should be decided whether the child should continue to be cared for at a CF centre or by the family doctor.

What does this mean and reasons for caution?

This new guidance on the follow-up of children with an inconclusive diagnosis will help the CF team to work in partnership with the family to determine future care arrangements and come to a shared decision on the best way forward, which may include discharge to primary care with appropriate information. Information is key for these families, and we recommend consideration of a further consultation when the individual is a young adult to directly communicate the implications of an inconclusive diagnosis.

What's next?

We would like to set up a registry for children with an inconclusive diagnosis to see what happens to these children in the future and how many of them convert to a CF diagnosis.

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