



Cystic Fibrosis Research News

Title:

Defining key outcomes to evaluate performance of newborn screening programmes for cystic fibrosis

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What was your research question?

To establish key outcomes to determine and compare the performance of Newborn screening programmes and define parameters needed to calculate those outcomes.

Why is this important?

To improve the collection of data and better enable comparison of the performance of different NBS programmes we established key outcomes and parameters required to calculate those outcomes.

What did you do?

The outcomes were determined to reflect four main algorithms identified in the last survey, but also aiming at being inclusive to new screening algorithms. A core panel of experts constructed a process map to determine key outcomes that would best reflect performance and illustrate the impact on the family and clinical progress. Once these outcomes were determined, all parameters required to calculate the outcomes were listed.

What did you find?

Eight key outcomes were established:

1. Coverage of the NBS programme
2. Proportion of samples taken on to second tier testing
3. Number of infants with a CF or unclear diagnosis
4. Number of carriers recognised
5. CF cases missed by NBS
6. The ability to correctly identify infants with CF
7. The ratio of CF infants to those with an unclear diagnosis
8. Age at initial visit at a CF centre

Definitions were established for each outcome and a panel of 20 parameters was developed to be collected to calculate these outcomes

What does this mean and reasons for caution?

The considerable variation of NBS programmes in Europe is not ideal, and clear outcomes are required to rigorously assess and compare the performance of different protocols. We have developed for the first time a number of outcomes with clear definitions, and we have also established parameters required to calculate the outcomes reliably. By generating a list of outcomes with clear definitions, the collection of data from countries and regions with NBS



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programmes will be more consistent, enabling better evaluation of performance. Through comparison of outcomes, programmes can adapt to achieve national standards and improve quality.

What's next?

We are planning a new survey on the performance of newborn screening programmes in Europe.

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