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Cystic Fibrosis Research News

Title:

Implementation of newborn screening for Cystic Fibrosis in Norway. Results from the first three years.

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What was your research question?

As of 2012, all newborns in Norway are offered Cystic Fibrosis (CF) screening in dried blood spots. The program aims to identify babies who have CF with a high degree of certainty. After three years, we evaluated how well the program performed.

Why is this important?

CF is one of the most common inherited life-limiting diseases in Caucasians. Both parents are healthy carriers of the genetic variants that cause the disease and the baby usually looks healthy at birth. Early symptoms are unspecific and often not recognized as CF. If left untreated, affected infants may become undernourished and suffer damage to lungs and other organs. When correct treatment is started early, the prognosis is dramatically improved.

What did you do?

All samples were tested for the CF marker Immunoreactive Trypsinogen (IRT). However, this marker is not specific enough to be used alone. Samples with raised IRT were therefore tested for genetic variants believed to cause CF. The variants found in people with CF in Norway, as well as a range of internationally known variants, were included in the test

panel. Only infants with two detected disease-causing variants were reported for diagnostic follow-up.

What did you find?

Surprisingly, a large group of infants reported from screening could not be definitely diagnosed with CF. All of these infants carried the same specific gene variant, which was not identified as common in our CF group prior to screening. There were also cases of infants with gene variants that had not previously been seen in Norwegians with CF.
One infant with low IRT developed symptoms of CF and was not detected by the program.

What does this mean and reasons for caution?

The aim of newborn screening is early detection of infants with CF. Healthy carriers of CF are not reported using our workflow. This strategy results in less worry for parents, but might miss out on rare variants that are not covered by the screening test. In order to detect as many affected infants as possible, a wider range of variants known to cause disease should be included.

What's next?

Variants that are re-evaluated as unlikely to cause CF will be removed from the panel.
We also wish to use new sequencing technologies, allowing detection of more variants with lower cost per sample. We could then lower our cut-off point for levels of IRT. Hopefully this will further improve the performance of the screening program.

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