



Cystic Fibrosis Research News

Title:

Normal pancreatic function and false-negative CF newborn screen in a child born to a mother taking CFTR modulator therapy during pregnancy.

Lay Title:

Infant with cystic fibrosis has normal pancreas function and was missed by newborn screening after mom stayed on CFTR modulator therapy during pregnancy.

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What was your research question?

If a mother takes CFTR modulators during pregnancy, does it affect a newborn who has cystic fibrosis? This is a case report, not a research study – the mother was already taking a CFTR modulator and decided to keep taking it for her own health during the pregnancy.

Why is this important?

A growing number of women with cystic fibrosis have started taking highly effective CFTR modulator therapies (HEMT). Not only has this improved the pulmonary health for these women, some are seeing improved reproductive health. They have been able to get pregnant and carry babies to term more easily. This is the first known case of a baby with CF being born to a mom taking HEMT. Unlike most babies with CF, a newborn screening blood test that detects pancreas problems was normal. Testing for CFTR mutations needed to be requested since there was no sign of pancreas problems.

What did you do?

Once we knew the baby had CF with two of the most common CFTR mutations (F508del), standard CF care guidelines were followed. A sweat chloride test confirmed CF, but the sweat was not as salty as it is for most infants with CF. We check stool samples each month to see if enough digestive enzymes are being released by the pancreas. We also checked for possible side effects from HEMT exposure by checking liver function and a detailed eye exam.

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What did you find?

So far, the baby is thriving and not showing any signs of CF except having salty sweat. There has been no problem gaining weight or growing. The elastase levels that were checked in stool samples to measure pancreas function have all been normal. The liver tests did not show any side effects of being exposed to HEMT before the baby was born. The eye exam was also normal, without any signs of cataracts.

What does this mean and reasons for caution?

Being exposed to HEMT before birth might prevent some CF-related organ damage for newborns who have CF. This has been studied in ferrets with CF, and treating them with HEMT during pregnancy leads to healthier baby ferrets with CF. Without HEMT after birth, those baby CF ferrets developed CF-related organ damage. Thus far, our patient has not shown any CF-related digestive problems even though there is no approved HEMT available to newborns. Much more study is needed before anyone would recommend HEMT treatment for pregnant women in order to protect a baby who has CF mutations.

What's next?

With more women who have CF and take HEMT having babies, it is possible that some of those babies will also have CF. It is important to check for CFTR mutations in the babies even if they have normal newborn screens.

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