

# Cystic Fibrosis Research News

**Title:**

Clinical outcomes of two infants with cystic fibrosis, including presence of the vas deferens, born to a woman with cystic fibrosis taking CFTR modulators during both pregnancies

**Lay Title:**

Presentation of two siblings with CF born to a mother with CF taking Trikafta (Kaftrio) throughout both pregnancies. A baby boy has normal vas deferens after birth.

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**What was your research question?**

This is a presentation of a woman with CF and her two babies with CF. The woman was treated with Trikafta through both pregnancies thus exposing both her babies to the treatment. We report how these babies with CF were affected by being treated with Trikafta before birth.

**Why is this important?**

There is a growing number of women with CF who are getting pregnant more easily on Trikafta after years of struggling with infertility. However, there is little known about how these babies are affected and if there are any negative effects. There is even less known if the babies have CF themselves. Our report suggests that if a baby with CF is treated with Trikafta before birth, the organs such as the pancreas and vas deferens, that are usually not functioning at birth, could be rescued and function normally.

**What did you do?**

We describe two siblings with CF born to a woman with CF. The woman started Trikafta due to low lung function (24 %) and got pregnant shortly after. She remained stable during pregnancy and delivered a baby girl at week 37 without complications. The genetic test after birth revealed that the girl had CF. After 6 months the woman became pregnant again. The amniocentesis showed that she was carrying a boy with CF. She delivered a baby boy at 36 weeks, this time by C-section due to early contractions. The infants were not breastfed due to the mother's anti-epileptic medication.



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## **What did you find?**

Both infants had normal pancreatic function after birth. The girl's pancreatic function decreased at 7 weeks, and she started taking digestive enzymes. She had her first iv treatment at the age of two. The boy had difficulty in gaining weight after birth and started on enzymes. He had a severe airway infection at the age of two weeks, which was treated with iv antibiotics. An ultrasound showed the presence of a normal vas deferens, which is known to be absent in male patients with CF. It also showed gallstones that are rare at an early age.

## **What does this mean and reasons for caution?**

Neither of the infants showed negative effects of Trikafta. The liver results and eye tests were normal. Both had normal pancreatic function suggesting that the pancreas could be rescued in babies with CF if they are treated before birth. The fact that the boy has a normal vas deferens means that he could possibly have children in the future. We do not know if the gallstones found in the boy, otherwise rarely observed in such young infants, are related to Trikafta or not. We share this experience to raise awareness among caregivers.

## **What's next?**

With more pregnant women with CF taking Trikafta, more infants with CF will be exposed to the treatment before birth. This experience shows possible positive effects of exposure to Trikafta before birth such as normal pancreas function and normal development of the vas deferens.

## **Original manuscript citation in PubMed**

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