Living with Cystic Fibrosis: A Mother's Journey of Strength and Hope

INTERVIEW

Cystic fibrosis (CF) is a complex genetic condition caused by mutations in the *CFTR* gene, resulting in persistent respiratory infections, digestive difficulties, and a significant impact on quality of life. While early diagnosis and medical advances have improved long-term outcomes, the daily reality of raising a child with CF remains profoundly emotionally and practically challenging.

In this deeply personal interview, we meet a remarkable mother living in the Netherlands, who is raising a spirited 6-year-old daughter with Cystic fibrosis, alongside her 2-year-old. She shares the joys, fears, and life-altering lessons that have shaped her family's journey. Through her candid reflections on diagnosis, treatment, and the demands of everyday life, she offers not only a glimpse into the realities of CF but also a message of strength, awareness, support, and hope for other families navigating similar paths.

How old was your daughter when she was diagnosed with CF?

I was about 20 weeks pregnant when the doctors saw something unusual in her intestines during an ultrasound screening. The doctors gave us a few options for what could have caused it, one of them being CF. We could have waited until she was born to find out the exact cause, using the heel prick test, but we decided to go forward with amniocentesis and know as soon as possible.

Can you describe your first thoughts and feelings upon receiving the diagnosis?

We were very worried about the state in which our daughter would be born and what her future life would look like. The scariest part was the thought of this disease taking her away from us way too soon.

Did you and your husband undergo genetic testing to confirm CFTR mutations? Were you offered any genetic counseling or support at the time?

Yes, we underwent genetic testing, which was a simple blood test. After the diagnosis, we immediately got a team of specialists to talk to.

What kind of medical or emotional guidance did you receive from these specialists when the diagnosis was confirmed?

There was and still is a whole CF team available to us at all times. This team includes pulmonologists, physiotherapists, dieticians, social workers, and psychotherapists specialized in children. We were also offered psychotherapy sessions for emotional guidance.

What were her early symptoms, and how have they evolved over time?

Our daughter got her medication from the beginning, so there weren't really any symptoms. With time, we noticed what most CF patients are struggling with, such as stomach pain or more condensed salt in her sweat.

Is she currently on any medications or therapies? If yes, when did her treatment begin?

Since birth, she has been taking enzymes which allow her to digest fat and vitamins A, D, E, and K, which she doesn't absorb well enough from food. For a while, she started taking extra fiber, which helps with digestion and stomach pain in general. Since she was 2 years old, she has been receiving medication (Orkambi), which is meant to improve her overall health. Soon she will start taking next-generation medication (Kaftrio), which is supposed to help with her condition even more. Our daughter's condition is under control and has been since she was born. However, I've heard from other CF patients who have really struggled with their health, taking lots of antibiotics and having low lung capacity that Kaftrio was a life changer for them. Additionally, for about a year, she has been doing almost daily inhalation with salt solution 7.5%.

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How often do you go for check-ups, and what do they include?

In the beginning, and for the first 2 years of her life, she had a check-up every 6 months. Doctors were checking her stomach, lungs, ears, eyes, taking a cough swab, and a blood test to check for many parameters. Since then, the check-up has become annual. In addition, the examination now includes checking her lung capacity and liver every 3 months to make sure the medication doesn't affect them.

Have you seen noticeable improvements or setbacks with her treatment plan?

Since she was born in quite good health and she's been getting medication and all the help she needs from the beginning, there have been no setbacks.

How has CF affected your family's daily routine?

I don't feel like it affected us much. We just need to remember to always have her medication with us and make time during the day for her inhalation. The only thing we've changed is getting rid of carpets, because of all the bacteria that are easily collected there. Also, we had to remove all plants from our house, as very dangerous bacteria for people with CF develop in the water and soil. Still, it would only affect her if she put her hands in it, so maybe we will slowly get some plants back as she gets older and understands more about how to follow hygiene precautions.

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What kind of support (e.g., from school, friends, or healthcare) has been most helpful?

I am very happy with the direct contact I have with the doctors and the nurses. If we have any concerns, they address them immediately. It's also very nice that our daughter's school teachers give her medication without any hesitation.

Are there any misconceptions about CF you've encountered that you'd like to correct?

Nowadays, it's not a death sentence anymore, and with proper care and proactive measures, you can have a mild version of the disease, and you can have a pretty much normal life.

What has been the most challenging part of this journey for you?

I think the hardest part was the unknown of what was coming. From the time of diagnosis until she was born, when we could finally find out definite answers about her condition.

How did the CF diagnosis influence your approach to your second pregnancy? Did you make any special preparations or undergo prenatal testing?

The chances of us having a child with CF is only 25%. But after it had happened already once, 25% sounded a lot scarier to us. That's when we decided to have a second child with an IVF procedure, to reduce the chances of our second child being affected by CF.

What would you want other parents, especially new or expecting parents, to know about CF?

The medical field for CF diagnosis is progressing rapidly. A lot has changed in the last 10 years CF-wise, and probably a lot will improve in the coming years. Be aware that the CF care might be slightly different in every country, so always keep your options open. Cherish every moment and remember that there is a whole community of parents like you, so you can always find someone to talk to and guide you through!

Disclaimers:

- *The content of this interview is intended for informational purposes only, and should not be perceived as medical advice. Always seek advice from your healthcare providers if you have questions or concerns about CF or treatment.*

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