

Complex Child E-Magazine

www.ComplexChild.com

Cystic Fibrosis? When Doctors Disagree on the Diagnosis

by Kim Gately

Hope, worry, frustration, and letdown. It's the cycle we face in our home every time Christopher sees another specialist or has an appointment with a doctor. For the past 12 years, it's been a vicious cycle for us, over and over and over again. We get so very close to a diagnosis, so close you can feel the relief wash over you and taste the thrill of finally being able to give what he has a name. Then fear and hopelessness set in as you realize your bubble has just burst and you're no closer to answers.

Christopher's Story

Christopher was born very healthy in July of 1994. He was a big, healthy eight pound, nine ounce baby with a good set of lungs and a vivacious appetite. Then, at age two, symptoms began. An asthma attack out of nowhere left him with blue lips as he was rushed to the ER. It was a feeling I never thought I'd have to cope with. It was followed with a diagnosis of asthma and allergies and a home nebulizer. Then, a few years later, it was pneumonia, ear infections and bronchitis that just wouldn't go away. We were told to get rid of our pets, remove the carpeting, and give him steroids. Things seemed to improve.

At age six it was an umbilical hernia that bothered him. The doctors said it would be a quick surgery and he would be fine. In truth, though, that was the beginning of it all. Surgery went well, but the asthma continued and then the constipation began. By age eight, Christopher was leaking stool on a daily basis and wasn't having regular bowel movements. No medications could get his body to regulate itself. He also stopped growing. He would gain a centimeter or a pound or two here and there, but he was slowly falling behind all his classmates in size.

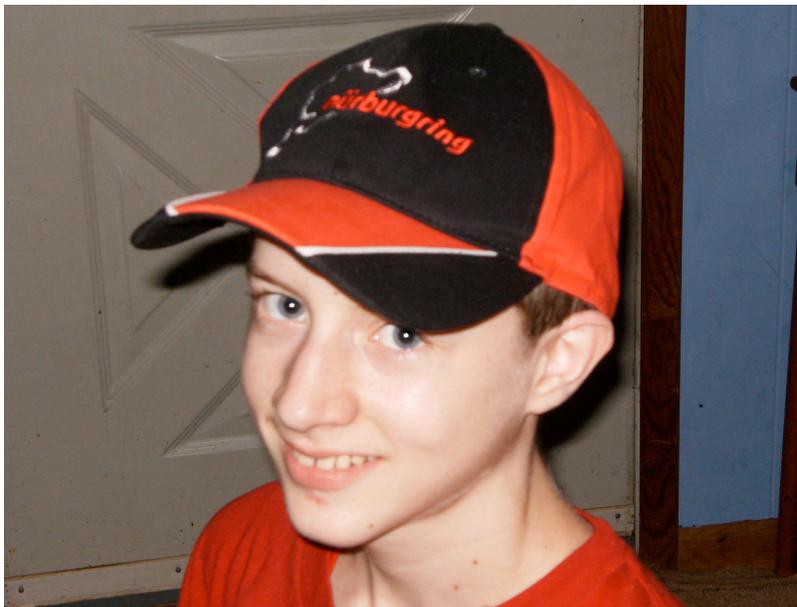
Finally, we saw a GI specialist. He swore to us that Christopher's constipation was laziness. "He's just too busy to take the time." Not true, I told him. He tries every day. Another doctor said Celiac Disease, a reaction to gluten in food, but the tests came back negative. Hirschsprungs? Nope, the biopsies were negative. Barium enemas, lower GIs, Sitz marker studies, colonoscopies and endoscopies only told us one thing: that he wasn't digesting food as he should, and after he did, it just sat there, stool building up until it had no choice but to come out.

Through all his GI problems, we still continued to battle his asthma, with attacks coming

unpredictably and with no known triggers. Finally, a hospitalization provided some relief in the form of a doctor who would actually LISTEN to me and believe me. Christopher's problems were not psychological or emotional. Something really was wrong. Our miracle doctor scheduled Christopher to see a motility specialist and we finally got our first answer: Colonic Neuropathy.

Toward a Diagnosis

The diagnosis of Colonic Neuropathy basically means that the nerves in the colon don't function properly. The treatment in a case as severe as Christopher's is a surgical procedure called an Antegrade Colonic Enema or ACE, which allows irrigation of the colon through a stoma or hole in his abdomen. He would have to face another surgery, but this one would finally bring a light at the end of the tunnel. It's permanent, and Christopher would never again have a bowel movement without irrigating his colon. But after years and years of frustration, it was so worth it as we had answers and a relatively simple fix. The relief was like nothing I had ever experienced. Surgery went smoothly and recovery time was minimal. Christopher was soon irrigating his colon like a champ and feeling better.



Relief was the new word in our house until the GI offered a disturbing idea: Cystic Fibrosis (CF). It can cause both the asthma and the poor growth Christopher has battled for years and explains the fatigue and weakness, too. We quickly scheduled an appointment at the CF center in our area. Further exploration into family history showed that Christopher's uncle, on his father's side, does indeed have CF. Finally, all the pieces started to make a puzzle. The outsides were together and we were just searching for that missing piece, the final diagnosis.

But when we met with the CF doctor, he performed a sweat test, a standard diagnostic

tool for CF, and it came back negative. This doctor was certain that Christopher did not have CF. Doubt still lingered in my mind, and with good reason. When I received the documentation, it appears the hospital didn't collect enough sweat to perform an accurate test, and thus the test was ruled incomplete.

Disagreeing Doctors

I made another appointment at a different CF center. We traveled two hours to get there and saw two different doctors. They also performed a sweat test and it came back borderline, ruled inconclusive. We saw the doctors separately. The first, a young doctor with great bedside manner, explained atypical CF to us. She felt that she could give us a diagnosis. Yes, it is CF, atypical, but CF none the less. I began to cry. Finally, we have answers and we can leave knowing our enemy and the battle we have to fight!

Then the second doctor came in. He felt that it was NOT CF, atypical or otherwise. I began to cry again, more with frustration than anything else. Why can't we just have answers? Why have we seen more than 15 doctors over the past eight years and not one of them can conclusively tell me what is wrong with my son?

We now have four medical opinions to digest. Two say yes, it's CF, and two say no, it's not. All we want is answers and now we know that the only way to be sure is to see a genetic specialist and get the necessary bloodwork done. That leaves us where we are today.

The Journey Continues

Christopher still suffers from asthma, scoliosis, weakness and slow growth. He also has headaches and knee pain that comes and goes. He has seen an orthopedic surgeon who is concerned about his bone structure in his chest and back, but the doctor found no underlying condition and couldn't give us answers. Christopher will soon see an ENT to decide whether or not they will operate to remove his nasal polyps and adenoids. His nasal drip is horrendous and he is completely unable to blow his nose.

Daily we fight to keep Christopher nourished, hydrated, comfortable and healthy. He is very often left out of normal teenage activities because of his medical problems. If he does participate, it often leaves him sick afterwards.

Our journey isn't over, by any means. We still seek answers and won't give up until we have them, but until we do, we just take it slowly and lovingly, taking life moment by moment, one precious day at a time.

Visit Christopher's website at <http://www.freewebs.com/for-cj>