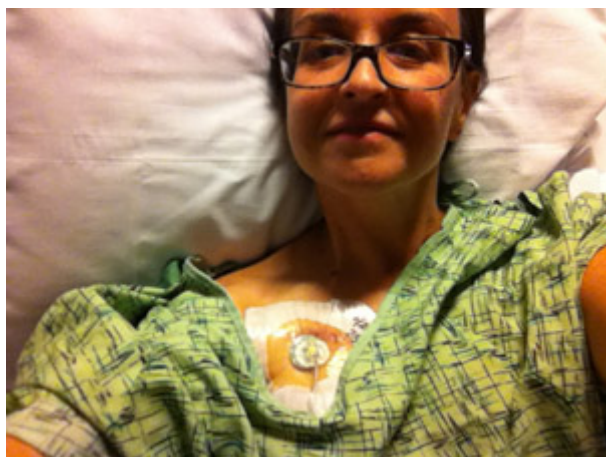


Mother Receives Novel Treatment for Rare Disease

By Michelle Brubaker | May 05, 2014

Ingrid is a working mother of two who has always led a healthy lifestyle. One day, she noticed her stomach was becoming upset after eating. At first, she didn't think it was anything serious, but weeks later, she was in horrific pain, had lost her sense of taste and her hair was falling out.



"After several tests and examinations, nobody could figure out what was going on with me," said Ingrid. "I had severe diarrhea and was running to the bathroom 15 times a day to throw up. I was so metabolically bankrupt and low on potassium, my doctors thought my heart would stop."

Although it mirrored similar symptoms, Crohn's disease was ruled out. Ingrid's medical condition continued to be a mystery even as she stopped eating to prevent pain and consequently lost 49 pounds.

Desperate for help, Ingrid was referred to [William Sandborn, MD](#), chief of the Division of Gastroenterology and director of the Inflammatory Bowel Disease Center at UC San Diego Health.

"Within two hours, I had an appointment and started to feel hopeful," said Ingrid. "I had to miss my kids' first day of school, but I was so sick I just needed to see Dr. Sandborn as soon as possible to survive."

When Ingrid arrived to Sandborn's office, she was incredibly malnourished and in a wheelchair.

"The examination room was full of fellows and residents studying this rare case," said Sandborn.

Tests results showed Ingrid's digestive tract was riddled with polyps. After further screenings and online research, Sandborn noticed Ingrid's fingernails were painted – a big clue to her diagnosis.

"When I asked Ingrid what her nails looked like under the polish, she told me they were yellow and brittle. I knew then she was suffering from Cronkhite-Canada Syndrome (CCS)," said Sandborn.

CCS is a very rare disease with symptoms that include loss of taste, overgrowth (hypertrophy) of the stomach and intestine, intestinal polyps, hair loss and nail growth problems. There have been fewer than 500 reported cases of CCS worldwide in the last 50 years. The condition has a 50 percent mortality rate, primarily due to severe malnutrition.

“Due to the hypertrophy of the stomach and intestine, patients with CCS have a decrease in their ability to properly absorb nutrients through the intestine,” said Sandborn.

The disease is not believed to have a genetic component but most often affects middle-aged male patients of Japanese descent.

“As an American woman in my 40’s, I certainly did not fit the profile,” said Ingrid.

The cause of CCS is unknown and the goal of treatment focuses on symptomatic management and nutritional support. Ingrid was soon treated with total parenteral nutrition (TPN) – a method that provides patients nutrition intravenously.

“Within 12 hours of TPN, I felt like a new person,” said Ingrid.

Once Ingrid’s body was more nourished, it was time to look into a novel way to put her disease in remission. Surgical removal of polyps would reduce the risk of cancer, but it wouldn’t relieve her nutrition problems.

“Through a multidisciplinary team effort, we decided to use Remicade to treat Ingrid. It’s a drug historically used to treat rheumatoid arthritis, Crohn’s disease and ulcerative colitis,” said Sandborn. “It’s an unusual use of Remicade, but we saw a dramatic improvement in Ingrid soon after she started taking the suppressive drug.”



Ingrid and her husband celebrate their 16th wedding anniversary.

Ingrid is now considered to be in remission but continues to receive Remicade via IV every eight weeks. The art teacher says the drug can make her a bit tired and suppresses her immune system, but she is more than willing to deal with the side effects.

“I am so grateful to Dr. Sandborn for not being afraid to try innovative approaches in medicine that saved my life and allowed me to return to my full mommy duties,” said Ingrid.

She has also shared her experience in a blog that has turned into an online support system and a way to connect with other patients around the world.

“My blog allows me to reach out to others so they don’t feel alone and to put my journey in perspective. My life has

become more meaningful, and I am grateful for every day," said Ingrid.

"It has been an honor to go through this journey with Ingrid and put the pieces to the puzzle together, which has resulted in her living a full life with her family," said Sandborn. "It's what I truly love about my job."

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