Heather Kirkwood

HERMANSKY-PUDLAK SYNDROME

"I made the nurse take a picture of me taking my first pill. For me, it was a grand moment. It was the moment I went from a gloomy certainty about what my future held, to a blissful land of the unknown."

I wear a lot of hats. I'm a daughter, sister, editor, patient advocate, and participant in the phase III clinical trial of Pirfenidone to treat pulmonary fibrosis of Hermansky-Pudlak Syndrome (HPS).

HPS is a rare type of albinism that causes vision impairment, bleeding disorder, and, in some mutations, digestive problems and ultimately pulmonary fibrosis. Although my lungs were relatively healthy, the road to becoming a clinical trial patient was a long one of misdiagnosis, misinformation, and the realization that I was not invincible—a shocker for a 29-year-old.

My background as a journalist compelled me to gather every piece of information I could, but none of it seemed good. I read abstract after abstract that stated, "usually fatal in the third to fourth decade." If they were really generous, they said fifth decade. Even worse than my diagnosis was having to tell my 25-year-old brother that we shared this diagnosis. I couldn't bear the idea of one of us watching the other die.

I became involved in the Hermansky-Pudlak Syndrome Network and contributed however I could, whether it was producing the newsletter, holding a fundraiser, or managing the online support communities.

And one day, fortune smiled on us. The National Institutes of Health was able to follow up their phase II Pirfenidone trial with a phase III trial. As a community we were beyond thrilled. But when I was screened, I was deemed too healthy to be admitted.

A year later I felt as though my asthma was getting worse. I just didn't seem to have the endurance I normally had walking around the neighborhood and running errands. I did my yearly pulmonary function tests at home and I was nervous.

HERMANSKY-PUDLAK SYNDROME

- Hermansky-Pudlak Syndrome is a genetic disorder that causes pulmonary fibrosis in certain subtypes.
- Pulmonary fibrosis occurs nearly 100 percent of the time in HPS gene types 1 and 4, although the age of onset varies.
- HPS is characterized by albinism, decreased visual acuity, nystagmus, a bleeding tendency, and, in some, a digestive problem similar to Crohn's disease.
- Genetic testing for HPS can result in false negatives because not all HPS genes have been identified.

Source: Hermansky-Pudlak Syndrome Network—www.HPSNetwork.org

I discovered that my FVC had fallen from 94 to 69 and my DLCO had fallen from 70 to 53. I reapplied to be included in the trial and was accepted.

I remember picking up my pills for the first time at the pharmacy at NIH. I looked at the bottle with my name, my patient number, and the words "Pirfenidone or placebo." I don't know which I'm receiving. My doctors don't know. Only some bureaucrat statistician in the depths of the NIH knows what's really in those bottles.

I made the nurse take a picture of me taking my first pill. It's a horrible picture of me and she seemed to think I was a little strange. But for me, it was a grand moment. It was the moment I went from a gloomy certainty about what my future held, to a blissful land of the unknown.

It was the day I got my hope back.

I know Pirfenidone isn't a cure. I know this is a trial. And I know I might be on a placebo. But it's something—it's a step forward.

It means that just maybe those horrible paper abstracts won't apply to me.

Heather Kirkwood was a patient speaker at the ATS 2009 International Conference in San Diego, Calif.