Carmen Camacho

HERMANSKY-PUDLAK SYNDROME

Carmen Camacho was a patient speaker at the ATS 2018 International Conference in San Diego, California.

My name is Carmen Camacho. I am 53 years old, I am legally blind, a mother of two, one of them with autism, and the main breadwinner at home in Massachusetts. I have a MA in psychobiology and work as a social worker doing clinical work with individuals with chronic mental and physical illness. I have pulmonary fibrosis from Hermansky Pudlak Syndrome, Type 1.

I may not look like your typical palliative care referral but I am, even though I have not had to deal with the intensity of the symptoms of the fibrosis. As part of my job as a social worker, I work with families waiting for transplants. Seven years ago, I was at an HPS client-turned-friend's bedside. She qualified for a transplant and there was a big storm in the Northeast. Her family was two hours away, so she asked me to stay with her until they got there. Her transplant coordinator came over to talk to me about the case, but then looked at the two of us and said, "Wow, this must be like looking in the mirror." I had never thought about that before, but it was. Since then I have seen over 25 of my fellow HPs'ers pass away.

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About four years ago I needed to have my ovaries removed due to the bleeding of HPS. At that time, I needed to get Human Leukocyte Antigen matched platelets which were really difficult to match because my plasma renin activity was so elevated. I didn’t think about it until I came to an ATS Patient Day. They were talking about factors that might disqualify a patient for a lung transplant, among them, antibodies. At the end of the talk, I approached the doctor about high PRA levels, and the chances for a lung transplant. He answered very candidly that anyone with a PRA as high as mine had been would be disqualified immediately. I just looked at Donna Appell, the president of the HPS network, and tears burst out of my eyes. Donna just held me and let me have my moment. I guess she was my palliative care team.

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Hermansky-Pudlak Syndrome

Hermansky-Pudlak Syndrome (HPS) is a rare inherited disease, named after two doctors in Czechoslovakia who, in 1959, recognized similar health conditions in two unrelated adults. Since the discovery of HPS, the condition has occurred all over the world but is most common in Puerto Rico. The most common health conditions associated with HPS are albinism, the tendency to bleed easily, and pulmonary fibrosis. A growing number of gene mutations have been identified causing HPS (including numbers HPS1 to HPS10). A breakdown of common issues HPS patients experience are:

- Albinism is an inherited condition that reduces pigmentation (coloring) present in the body. People with albinism are often with fair-skinned with light hair. Low vision and carious degrees of nystagmus is seen in all cases.
- HPS patients have normal numbers of platelets, but they are not made correctly and do not function well, so the blood does not clot properly. As such, persons with HPS may bruise easily or have frequent nose bleeds.
- The exact cause of pulmonary fibrosis in HPS patients is uncertain. However, there is inflammation present and over time the lungs become scarred which limits the ability for oxygen to enter the blood.

Learn more: ATS Patient Education Series What is Hermansky-Pudlak Syndrome? New York, NY. ***www.thoracic.org/patients/patient-resources/resources/hermansky-pudlak-syndrome.pdf***