Candice and Crystal Sipe

HERMANSKY-PUDLAK SYNDROME

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We are identical twins, adopted at the age of one from Hyderabad, India. It might surprise you that we are Eastern Indian, but we have albinism because of Hermansky-Pudlak Syndrome type 4. HPS is a genetic metabolic disorder. It causes legal blindness, albinism, platelet dysfunction and in our case, inflammatory bowel disease, severe osteoporosis, acid reflux, kidney disease, and pulmonary fibrosis. Approximately 1,200 people are on the HPS patient registry, and there is no cure at this time.

As babies we were very white with blond curls, and our eyes were in constant movement due to the nystagmus. At 15 months old, we saw a pediatric ophthalmologist and were fitted with glasses. We both had eye surgery a few years later to help slow down the nystagmus. If we had known about HPS at the time, we would have held off on surgery because bleeding was a considerable issue, and we both bled profusely.

We can't think of a time in our lives when we weren't battling a health challenge. We saw many doctors who made various diagnoses. As children we dealt with many asthma attacks and constant difficulty breathing. Now as 34-year-old women, our asthma has been well controlled with the help of steroid and rescue inhalers. We both have had upper respiratory infections, including pneumonia and bronchitis. Candice has had Valley Fever, and she was on treatment for many years.

We have seen several pulmonologists throughout the years. Since HPS is so rare, most doctors don't know about it. We have to educate them, and very few are willing to research and learn about the disease. In the process, we have become our own advocates and researchers. We have chest CT scans every other year and pulmonary function tests regularly (our FVC is at 60 percent). I am thankful to say neither one of us has

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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 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).

- HPS gene causes pulmonary fibrosis in gene types 1, 2 and 4; therefore, it can be followed prior to patient reported symptoms.
- HPS is characterized by a platelet bleeding disorder. In the past, the standard of care supported platelet transfusions to prevent bleeding during surgical and dental procedures. Because of the formation of antibodies that can cause difficulty matching lungs for transplant, the recommendation is to keep platelets on standby until needed rather than transfusing liberally.
- Postoperatively, platelets and medications that enhance clotting are routinely prescribed after surgeries. Careful use of this therapy with attention to dosing and timing is necessary as DVT's and unwanted clotting have occurred from possible overcorrection.

Learn more: ATS Patient Information Series. "What is Hermansky-Pudlak Syndrome?" New York, NY: American Thoracic Society 2015. thoracic.org/patients/patient-resources/resources/hermansky-pudlak-syndrome.pdf.

pulmonary fibrosis yet, but we know it could happen at any time because of our age.

Singing is our love and our passion. We take zumba classes three times a week. Physical activity is tough, but we have been very faithful with our exercising and both feel that staying active helps keep our lungs strong. It's important for us to stay as active as we possibly can.

At age 12, I finally saw my first pediatric GI specialist, who was concerned that I wasn't responding to the medications like a normal Crohn's patient should. He sent our family to Cedars-Sinai Medical Center, where we were diagnosed with HPS. Our family was devastated to learn this news, and we had no idea what HPS was, or where to begin. Our doctor told us that he knew of a colleague in New York with an HPS patient. The patient's mother was Donna Appell, founder and president of the HPS Network. We were so overjoyed to find the network; it was as if a huge burden was lifted from us. We were no longer alone.

Meeting Donna allowed us the opportunity to get to know our HPS family. In November 1995, Crystal and I were invited to open the HPS protocol for research at the National Institute of Health. Accompanying us was Donna's daughter, Ashley. Our team is wonderful, and we continue to follow up with one another regularly. We are thankful to have the support of such a tight-knit group of people.