“We were so overjoyed to find the network; it was as if a huge burden was lifted from us. We were no longer alone.”
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
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.