

Leslie Rojas

Leslie Rojas of Puerto Rico, has Hermansky-Pudlak syndrome and needs to move away from her home, friends, and family to have a chance to enter a transplant program on the mainland.



My name is Leslie, and I was born with a rare genetic disease. I have no pigmentation on my skin. I have lived my whole life in Puerto Rico, where the sun's UV index runs from moderate to extreme all year long. I did not have access to sunscreen. Throughout my childhood my parents tried to keep me indoors to prevent my skin from burning, but I always found a way to go out and play. When I started primary school, I learned I was legally blind.

One afternoon, when I was 14 years old and visiting my aunt in New York, her dog chased me around the apartment and I fell. When I stood up, my face was covered in blood. The bleeding did not stop, so I was rushed to the hospital where I continued losing blood through my nose and mouth for about five hours. I was admitted to the hospital that night, but I didn't understand any English so I could not understand what was happening. I was hospitalized for a week and after multiple tests, doctors diagnosed me with Hermansky Pudlak Syndrome (HPS).

After completing college, I began searching for a job as a chemist. It was almost impossible for a legally blind chemist to find a job. A year passed, and I was offered a position as a laboratory technician at a pharmaceutical company. I had to work with solvents that in some instances could be dangerous to the lungs, so every year they would perform a pulmonary function test (PFT) on the lab analysts. I still did not know about the lung element of HPS. When they found that my lung capacity was declining every year, I was referred to a pulmonologist. I had a Gallium scan and the results of the test indicated that I had an inflammatory process consistent with pulmonary fibrosis (PF). The results were devastating. I had a terminal disease and there was no treatment, no cure, no hope.

I was so scared! For days I could not sleep, nor eat a full meal. I could not think rationally and could not go to work, where I had been relocated out of the lab, and

had started working on computer systems. At nights I just laid down on the sofa crying until I fell asleep, just to wake up an hour later to start crying once again. I remember trembling and shaking, and my teeth chattering uncontrollably. In time, accepted my diagnosis and decided to learn more about my condition. I visited several pulmonologists in Puerto Rico, but none of them could agree on the stage of the pulmonary disease, or on a treatment option. One of them mentioned that the NIH had a protocol on HPS related pulmonary fibrosis, but I would not be accepted because I had HPS Type 1, which he said was “the less severe manifestation of HPS.” I tried to explain to him that Type 1 was the most serious form of the disorder, but he told me I was wrong and did not provide the referral to the NIH. Little did he know that the NIH was only a few clicks away from an educated and persevering patient.

I contacted the NIH directly and I began a drug trial with the NIH in 2006. I was on the medication for three years and was stable up until 2010, when they first started to see some scarring on my lungs. In 2017, we started seeing a major change from my annual CT scans, and that’s when the doctor told me I should move to the mainland U.S. so I could be close to a hospital where I can have a lung transplant. Hospitals in Puerto Rico perform kidney, liver and even heart transplants, but no there is no lung transplant



center on the island, so I would have to leave my job, my home, friends, and family to have a chance to enter a transplant program.

I started researching hospitals recommended by my pulmonologist at the NIH, but then we were hit by Hurricane Maria. I lost the connections I’d made, and so I went to stay with my sister in New Jersey. From there, I traveled to Boston and had my first evaluation for a transplant. It was an interview, not a medical evaluation, but the doctor told me I was not ready to enter into the lung transplant center because I was

too healthy. I returned to Puerto Rico, and I talked to the NIH. They went back to the doctors in Boston and told me I should go back and start on relocation, but since I was already back in Puerto Rico and feeling well, I decided to stay.

I decided to go back for evaluation last year, so in September of 2019, I went to Boston to begin the series of tests to prepare for the transplant. I told them that I had a thyroid nodule, and I brought all my x-rays and ultrasounds. I also scheduled an appointment with a surgeon in Boston who told me that the nodule should normally be removed, but because I have a history of bleeding, I should first ask the transplant program. The



doctor in the transplant program agreed that the nodule should be removed and tested for cancer, because you have to be free of cancer for five years before you can even be considered for a transplant. I decided to return home to coordinate coverage with my insurance and coordinate after care with my endocrinology doctor. I also needed the results from an HLA platelet match test previously performed at the NIH, so my doctors in Boston could find a donor with the same antibodies to avoid rejection after the lung transplant. The thyroid surgery itself has to be performed in Boston, because I saw two experienced surgeons here in Puerto Rico, two of the best on the island, and neither will do the surgery because I am high risk for both bleeding and for the extubation process. I scheduled the removal surgery for March 23, 2020.

The COVID-19 pandemic changed my plans. My procedure was cancelled, and I could not travel. My pulmonologist recommended that I should stay home to prevent exposure to the virus and on March 13, I began to work from home. Since then, I have stayed at home working, ordering groceries with an app, and praying for the end of the pandemic. I have not visited my doctors and have not been able to schedule the surgery. This summer has been warmer than previous years, and we have received clouds of dust from the Sahara Desert that have resulted in many days of unhealthy air quality. The island has also been skimmed by two tropical storms and earthquakes can be felt from



time to time. It seems that I will not be able to travel until a vaccine is available. I can only hope that my thyroid nodule stops growing, and the pulmonary fibrosis does not progress to a point where I would need oxygen, especially as we continue to lose power on the island as a result of the storms and earthquakes.

At this point, I am not currently on the transplant list because I still need to have the nodule removed. I need to have that nodule removed as soon as possible, before it turns to something bad, because so far tests have said it is negative for cancer but the last one was undetermined. I will not be on the list if the nodule is positive for cancer. That will be a big problem for me. ■

Hermansky-Pudlak Syndrome (HPS)

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 often seen in Puerto Rico. The most common health conditions with HPS are albinism, the tendency to bleed easily, and pulmonary fibrosis. Some other facts about HPS are:

- Albinism is an inherited condition in which reduced pigmentation (coloring) is present in the body. As a result, people with albinism are often fair-skinned with light hair.
- HPS patients have platelets that are not made correctly and do not function well, so the blood does not clot properly. As such, persons with HPS may bruise easily and have other issues such as frequent or heavy nose bleeds.
- Pulmonary fibrosis in HPS occurs in those individuals with HPS1, HPS2 and HPS4.
- HPS patients have reduced eye pigment and during early eye development the eye and surrounding nerves grow abnormally. This can lead to poor vision, the development of crossed-eyes (strabismus), and nystagmus (uncontrolled eye movement from side to side).



Learn more

ATS Patient Education Series.
"What is Hermansky-Pudlak Syndrome?" New York, NY.

<https://www.thoracic.org/patients/patient-resources/resources/hermansky-pudlak-syndrome.pdf>