



Donna Appell

Hermansky-Pudlak Syndrome

Our caregiving journey began like most, with the birth of our first child.

When Ashley was born, we were surprised by her full head of white hair. We discovered at her two-week “well baby” visit that she had albinism. That was the last time we had a “well-baby” anything. Christmas Eve of that year, we were told that our daughter was legally blind. I remember it how upsetting it was to think that your child was never going to be an astronaut, though I imagine that there aren't that many people who become astronauts. I wondered how she would do in school, and whether she would ever be able to drive.



Ashley's issue of low vision consumed us until we started to notice the bruising. I couldn't understand where they were coming from. I brought her back and forth to the pediatrician four times and was totally dissatisfied with his explanations. Eventually, I called the author of a pamphlet on albinism that I was reading because it mentioned a platelet defect. This expert sent me a test tube in the mail, and we drew Ashley's blood and sent it to him. He told us she had a disease called Hermansky-Pudlak Syndrome. I wanted to move to his neighborhood in Minnesota and have him be her doctor! We didn't move, but he guided us in learning about this disease.

When she was two years old, she hemorrhaged to shock from the inflammatory bowel disease of HPS. I raced her to the hospital. She was there for three months, and got 36 units of platelets and 6 units of blood. She lost so much blood that she had a traumatic brain injury from lack of oxygen.

After that hospitalization, my husband and I decided that no family should ever be alone in this again, so we started the Hermansky-Pudlak Syndrome Network. We began to research HPS and discovered that many individuals with HPS die of a lung disease called pulmonary fibrosis in their young adulthood. That was Ashley's type of HPS, and her genetic fate.

Care for our daughter was not going to have an end date until the unthinkable.

I think the hardest part of caregiving is the feeling of helplessness, that loss of control. No matter how hard you try to make things better, the disease just progresses relentlessly. I'm not a researcher or a pharmaceutical company, or a physician with a prescription pad. I can't create a treatment or change the course of things. But I could teach my daughter how to cope with her medical challenges, or at least I can be a positive influence. That was something in my control. So, my caregiving responsibilities were not only just which doctors or specialists to take her to, or what medications to give her but how I could help her cope.

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First and foremost, people model what they see. I tried very hard to remain composed and calm, and that helped everyone to stay more relaxed. I worked actively, not passively, on teaching coping mechanisms as part of caregiving. I felt that as much as the doctor’s work on her body, my caregiving needed to work on her spirit. I thought of all sorts of ways to fill her “coping toolbox.” Distraction of course was high on the list. We would often engage in positive imagery. When she was having a procedure or having blood drawn, we would talk about a vacation trip or a happy memory. We had little reward system for her bravery during IV insertions. She would get a little wrapped gift, a reward, a trophy, if she would keep her arm still. These gifts were small and certainly not elaborate. For a while, her gifts were colorful combs! She built up quite a large comb collection because she was so ill with so many IV’s. It became a trophy of how brave she was.

As life went on, I realized how much time we were spending in the waiting rooms of doctors' offices. That was precious time to plan happiness! I would be packed and ready with a new book to read to her. As she got older, I packed puzzles to work on and games to play. Now that she's an adult, we have done yoga in the doctor's office while we're waiting, we have rearranged office furniture to do mailings, and we have been known to dance and have a sing-along! Life is an adventure, and it was my job to make it fun. To cope with the harshness of health challenges I felt it was important to see different perspectives. Things could always be worse. So, at a young age I had her support others with difficulties worse than hers. Volunteering to help others is a sure way to stop focusing on yourself. It is a proven coping mechanism. That "attitude of gratitude" also helps grow positivity. When we leave the hospital or the infusion center, where she has received an IV infusion every five weeks for the last 21 years, I pull my car off the road and have her applaud and clap for all the doctors and nurses that got her back on the road again! Providing hope was also high on my agenda. I would look for opportunities to inform her of all the people working on trying to cure the disease. I gratefully brought her to the ATS International Conferences to see how many researchers dedicated their lives to lung health. The "scenes of science" at the ATS is so uplifting it is like medicine itself.

I'm not sure if anything I did or still do really makes a difference for her, but I do know that my daughter teaches me resilience, kindness, and strength every day of her precious life. I am honored to be caregiver, but caregivers need support too, particularly when providing care to adults.

Though it was not the intent of the privacy laws to make care giving so difficult, its interpretation has been tremendously variable.

This difficulty with “caregiving” is growing with a “new” population of complex medical young adults surviving childhood with diseases that would have previously been terminal in childhood, and older adults living longer and longer. While obtaining Guardianship is one answer, it will not be accessible to everyone. A Health Care Proxy allows a person to appoint another person to make health care decisions only at the time they become incapacitated and incapable. There needs to be something accessible in the middle. The Enhanced Health Care Proxy would enable the signer to designate someone that could speak on their behalf immediately upon signing rather than at the moment that they became incapacitated. Co-signed by a physician, the Enhanced Health Care Proxy would allow an adult to have a caregiver involved and there would be no HIPPA violation.

In 2011 I brought this idea to our State Legislators and a bill was created. It had multiple sponsors in the Senate and the Assembly and was referred to the Committee on Health. It was fully supported by the NY Nurses Association. It fell by the wayside because I was too busy “giving care” to lobby further for caregiving. ●

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. 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.

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