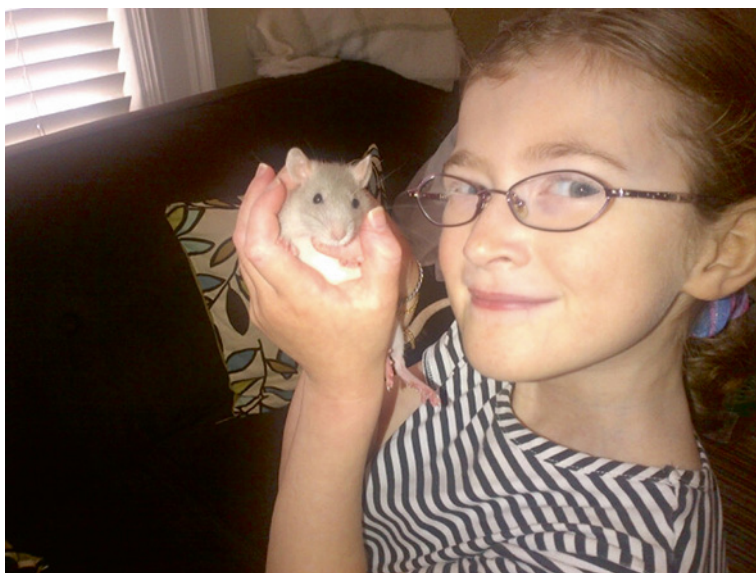


Vignette: Autoimmune Polyendocrine Syndrome Type I (APS 1)

A.K.A. Autoimmune Polyendocrinopathy-Candidiasis-Ectodermal Dystrophy (APECED)

Patrice F. Band



Welcome to APS 1: An Exclusive Group

Our only child, Julia, suffers from an orphan disease known as APS 1. It is an autoimmune disease that causes dysfunction predominantly, but not exclusively, in the endocrine system. It is a genetic disorder associated with a defect

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in the *AIRE* gene. Currently, it is believed that APS 1 attacks one in 2,000,000 people: approximately 17 in Canada, and 160 or so in the United States. While it can attack many tissues in the body, it is best known as a disease that causes hypoparathyroidism (leading to hypocalcaemia), chronic candidiasis (inability to fight off yeast infections) and adrenal failure (by itself known as “Addison’s Disease”). My wife Jennifer and I refer to those symptoms as the *trifecta*. But they represent only the minimum—the entrance criteria for membership in this small group. According to Julia’s doctors at SickKids Hospital in Toronto, and our own ever-expanding understanding, she was hit by the disease very early and very hard.

The Long Road to Diagnosis

Julia enjoyed a very healthy first year of life and met all of her physical milestones early. Shortly after her first birthday, she began to suffer from fevers that continued for days and a strange rash that looked like a map of the world (later identified as giant urticaria). After a bout of pneumonia and gastro-intestinal blockages, tests showed that Julia had dysphagia (a problem swallowing that can lead to aspiration of food and fluids). She then got sicker. From that point, she lived the better part of her toddler-hood in hospital, undiagnosed until she was about two-and-a-half years old. She spent almost three months in the CCU (Critical Care Unit) and most of that on a ventilator due to serious lung infections related to chronic aspiration.

She was seen by scores of talented specialists at SickKids, a smaller group at the NIH in Bethesda, MD and many were consulted world-wide. None knew the answer. As one senior pediatrician told us in the early days, “sometimes, we need to wait and the disease will present itself”. In Julia’s case, APS 1 arrived wearing a disguise. Chronic lung infections, urticaria, prolonged fevers, gastro-intestinal blockages and oral thrush are not specific to APS 1. So it was not until hypoparathyroidism emerged and genetic tests were ordered that the diagnosis was made.

During that year-and-a-half, Julia was followed by over 10 departments at SickKids. Leukemia, lymphoma and degenerative muscle disease featured on the differential diagnosis. It was a terrifying period of limbo. As disease after disease was ruled out, APS 1 caused serious and irreparable damage to Julia’s lungs, parathyroid, adrenal, salivary and tear glands. From this, further complications have cascaded.

To our surprise (and eternal gratitude), it was Julia’s dermatologist who first mentioned the possible diagnosis. *Autoimmune Polyendocrinopathy Type I*. I asked her to repeat it. I wrote it down on a yellow sticky as though it was the winning number of the next lottery. And I kept it until the genetic results came back.

Such is the hunger for diagnosis. Any diagnosis.

Treatment at Last

After the diagnosis was made, we agreed to an aggressive trial of steroids. The results were dramatic and the long road to rehabilitation began. Now, almost seven years later, Julia's complicated medical regimen includes immune-suppression therapy and supplements, and she still has a g-tube because her dysphagia has improved but has not yet resolved. While she is frequently sick and often in hospital, she is otherwise a thriving, beautiful, bright and active girl. She is also wise beyond her years and her spirit is an intoxicating force. But most importantly, she is happy.

What I Have Learned...

Patient Care is like Warfare

Over the course of caring for Julia through the various stages of her illness, from undiagnosed and grave to identified and chronic, I have learned that patient care is like warfare. Some battles occur, of course, between the patient or her caregivers and the doctors and hospitals, but that is not what I mean. What I mean is that there is a common enemy—disease—against which many people fight using the various tools at their disposal. This war is waged by patients, their families, caregivers and countless healthcare professionals with differing backgrounds over many weeks, months and years. It is waged with high-tech weaponry: modern diagnostic instruments, pharmaceutical products and non-traditional supplements. In response, the disease engages in covert and guerrilla tactics.

As a caregiver, one has to engage in relentless efforts at advocacy, diplomacy and bargaining—usually in combination. And as with any lengthy war, battle fatigue can set in. Some become separated, never to be heard from again. Many stop fighting: some because they do not understand the cause, others simply from exhaustion. In this, caregivers have to focus their dwindling energy on ensuring that the goal is kept in sight.

Lost Health must be Mourned

Anyone who has been touched by serious illness understands intimately that lost health, like other bereavements, must be mourned. Rare diseases can take more time to diagnose than more common diseases because medicine tends to operate on the principle that “common things are common”. Diagnosis is a process of elimination. So, in relative terms, the patient can lose more to a rare disease

than to a readily discernible one. In the case of some rare diseases, the hardship of mourning the former “normal” self is compounded by the awareness that some health could have been salvaged with early diagnosis.

Rare Diseases can Cause Extreme Isolation

Serious illnesses isolate people. Julia has spent a significant part of her life in hospital, away from her peers, and so have we as parents at her bedside. Because she routinely suffers from pneumonia, she must frequently be kept in “isolation” within the hospital for infection control purposes. When that happens, she is kept separate from her peers in the community *and* in the hospital. At times like those, we are especially thankful to have Tory, Julia’s loving COPE Service dog.

SickKids is, by any standard, a lovely building with fountains, several restaurants, and even a shopping area. But when you live there for months, it loses its veneer as you see the same exhausted parents shuffling up and down the halls, coffee in hand, around the clock. At those times, I have compared it to the island of misfit toys.

Disease also isolates people by making them (and their caregivers) different, or “other”. Even when not physically isolated, you can often feel alone because family, friends and coworkers simply do not (and cannot) understand what you are going through. While it is almost an oxymoron, isolation can be felt together, as a couple or family. Fortunately, there are groups of people who band together under the banner of their particular disease. I always knew these groups existed in the world. They sold lottery tickets and marathons were devoted to them. But they did not exist in my world. They do now. For rare diseases, though, they are harder to find and their populations are both smaller and more diffuse. They are therefore less powerful at attracting attention and much needed research funds than their more “mainstream” counterparts.

We in the orphan disease camp can feel connected to one another on the internet, but we are literally countries apart in the real world. Nevertheless, these groups are an enormous wealth of knowledge. They empower us to bring information to the attention of health professionals. They also give us a sense of community, albeit on a virtual island of misfit toys.

Adapting to a New and Ever-Changing Normal

Sufferers of serious disease and their caregivers have to adapt to a “new normal” in ways that can be unimaginable to the healthy. Some are unable to. We have seen families permanently torn apart by life in the SickKids CCU. Others weathered the storm together, only to fall apart when relative calm was restored. In our case, APS 1 has tightened the bonds of our small and extended family.

APS 1 is poorly understood and its course is difficult to predict. It took hold of Julia's life when she was one year old—among the earliest recorded cases, we are told. No one can explain why Julia, who is cognitively normal (gifted, in fact), has dysphagia. Every time she has pneumonia, her team wonders whether it is infectious or auto-immune. Many of her high and countless fevers strike without warning and leave the same way. Others foreshadow hospitalization. APS 1 is also associated with other serious illnesses including autoimmune hepatitis, retinal disease and diabetes, to name just a few.

So for us, living in the “new normal” is a bit like walking through a minefield. But we do it hand-in-hand-in-hand. While Jennifer and I are in a constant state of “high-alert”, Julia is miles ahead of us. She loves to hear idiosyncratic things about herself as a baby. She was born on her due date. She did not sleep that night. She refused to be swaddled and had to have at least one free arm. Recently, she said of those things that “maybe I was trying to tell you and Mama that I had a rare disease”. I think Darwin himself would wonder at Julia's adaptability!

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