

## **A Medical Mystery**

Could Adult Polygucosan Body Disease (APBD) be affecting *your* family?

A medical mystery started in my family in the late 1970s. My mom was a fun-loving, gregarious woman who grew up in a very large Jewish family, with nine kids. Her family was one of the relatively few Jewish families in Fort Smith, Arkansas. Although she had the normal dose of health concerns up to age 50, she was generally in pretty good health.

Somewhere around age 50 she started complaining about bladder control. She, and her doctors, attributed it to “female” problems. She attempted to cope by using pads to keep her clothes dry and various products to protect the bed at night. Her trips to a urologist for painful urinary tract infections were helpful but did not provide any permanent relief. This condition slowly got worse over time.

By her late 50s Mom was complaining about “weak ankles.” She started wearing very thick socks, because they “felt better” on her feet. Another doctor, this time a neurologist, was added to the mix. As bad as the numbness, stumbling, and foot dragging became, mom struggled to stay on her feet as long as possible. This was a woman who loved taking long walks outside in the cool evening air. Giving this up was not so easy! She acceded to a wheelchair for the first time when a caring cab driver refused to let her off at a major airport until she agreed to let him call for Skycap assistance.

And then there was the sleeping. Mom couldn’t stay awake past 8:30 or 9 p.m., even when she’d had a morning *and* afternoon nap. She was still in her early 60s.

### **The mystery deepens.....**

**A**shkenazi Jew

**P**eripheral numbness

**B**ladder incontinence

**D**ecreased energy

Remember this combination of letters: **APBD**. It has now come to mean a lot more to me, my family, and many others. For another meaning of APBD, keep on reading.

As Mom sank, Dad continued his job as a high-level engineering manager with the public agency responsible for the design and construction of the Washington, D.C., subway system. In addition to his job and hour-long commute each way, he took on the cooking, shopping, and laundry. At the same time, he was dealing with a prostate issue that left him with his own urinary troubles. And yes, he would sometimes say that he had been so tired at work that he had fallen asleep at his desk. But he never complained about life’s many twists and turns. He and Mom had met at Yom Kippur services while he was stationed at Camp Chaffee in Fort Smith. Soon after, they fell in love, marrying right before Dad shipped out to fight in World War II. At age 63, he retired to take care of Mom on a full-time basis. They

were both 67 when Mom died in 1988. The funeral took place within three days. In the standard Jewish tradition, there was no autopsy.

After Mom's death, Dad was finally willing to look into his own medical needs. Very quickly, a urologist performed prostate surgery. It didn't do the job; Dad's incontinence stubbornly refused to improve. During his rehabilitation, it became obvious that Dad wasn't walking well. When Mom was alive, he'd been pushing her wheelchair, and using it for support. The bottoms of his feet had become numb, but the rest of the family never picked up on his mobility problem. After lots of testing, including MRIs and more, a neurologist performed spinal surgery to fix a "pinched nerve" in his neck. This had little or no impact on the numbness in his feet. In addition, even though he was no longer working and no longer taking care of Mom, Dad was still falling asleep multiple times a day. We had not even begun to put the pieces together.

But now looking back on it: **Ashkenazi Jew. Peripheral numbness. Bladder Incontinence. Decreased energy.** You've heard that before.

### **Some clues**

The similarity of symptoms between the two of them was a puzzle to us. We wondered if there had maybe been some kind of environmental contamination. Maybe something was in the well water they'd used before city pipes had been installed in their Columbia, Maryland, neighborhood. Was it something related to the heating and air conditioning business that Dad and Mom ran in Philadelphia when they were first married? We even wondered if there was something in the pickle barrel that Mom used to make her wonderful sour tomatoes and pickles.

Dad died in 1996 at age 75. This time, despite the religious prohibition, I asked for, and had an autopsy performed. We needed to know what, if anything was going on in my family.

More than two months went by until the results came in: A pathologist at the Oregon Health Sciences University had discovered microscopic particles in nerve cells throughout Dad's body. He called these particles "polyglucosan bodies." Dad's disease, the pathologist said, was "quite rare" and called Adult Polyglucosan Body Disease (APBD). He said the cause was unknown.

Over the next dozen or so years, dogged researchers in Israel, New York City, Toronto, Dallas, and elsewhere have been at work in their labs, and progress has been made to understand—and hopefully treat—this disorder. Research going on right now gives us hope that there will be treatments ready for testing in coming years.

### **Solving the mystery**

Now detectable via a blood and/or saliva test, APBD is a genetic disease that predominantly afflicts Ashkenazi Jews. It occurs in men and women as early as age 35. Early symptoms vary too, but almost always include peripheral numbness, diminished bladder control, and fatigue. Very often, medical

caregivers mistake the array of symptoms for MS, ALS, prostate problems, and a variety of other disorders. At this time about 200 people have been identified as having APBD—and the numbers are growing as the word gets out.

I've had the "pleasure" of learning much more about APBD in the past two years, since I was diagnosed myself. At age 60 I started to have some numbness in my left foot. Together with an increase in urinary frequency and serious fatigue, I had all the clues I needed to get myself tested.

Since then I have gotten involved in clinical studies and gotten to know others involved in the APBD research community. In the past two years I have met a number of other individuals affected by APBD. Their stories of the long path through multiple doctors, incorrect diagnoses, unnecessary treatments, and unwarranted surgeries are heartbreaking. Many of them have gone a decade trying to find a solution to a number of worsening symptoms. (The fortunate side of my parents' experience was that it allowed me to find out what was affecting my health without going through years of unnecessary doctors, treatments, and surgeries.)

The fact is that today most doctors, including neurologists, have never heard of APBD. A recent study by Dr. Marvin Natowicz of the Cleveland Clinic indicates that 1 in 34 Ashkenazi Jews are carriers. The APBD Research Foundation is launching a campaign to help assure proper diagnosis of this disease. Countless surgeries could be prevented. Countless prescriptions for inappropriate medication could be avoided.

An APBD Patient Registry is now open that allows maintenance of a secure database of patients who might be good candidates for testing of new treatments as they become available. But only if properly diagnosed can patients participate in these drug trials—and get proper help for a devastating disease.

If you or someone you know exhibits symptoms of APBD, please consider getting tested. For more information please visit [www.apbdrf.org](http://www.apbdrf.org) and if you have any questions write the APBD Research Foundation at [info@apbdrf.org](mailto:info@apbdrf.org).