

# Real People Real Urgency

**Making an Early Diagnosis  
in Rare Diseases Can Make  
a Meaningful Difference**



## **Diagnosis:**

Late-Onset Sandhoff Disease

## **Symptoms first noticed:**

Eight years old

## **Diagnosis Received:**

Forty-eight years old

### **How were you diagnosed?**

Genetic testing.

### **How old were you at the time symptoms began?**

At 8 years of age, I would have tingling sensations in my hands and feet after physical activity and when I was not feeling well. The sensation was a lot worse than when my hands and feet just fell asleep.

### **What were the signs that triggered concern?**

At age 22, I started to notice leg weakness when using stairs or getting up from a seated position.

### **Did you share your concerns with your doctor?**

I shared my concerns with several doctors, including two neurologists. The first doctor told me it was all in my head, and the second doctor just didn't do anything. I went to a neurologist about a numb spot on my left leg and he thought it was merely a pinched nerve.

### **What led to the diagnosis?**

After so many doctors, I gave up on them and myself. After about ten years, a friend made an appointment for me without my knowledge and I agreed to go. The first thing I told the doctor was that I have trouble standing up from a chair, going up steps, and getting up from the floor. We talked a while and he referred me to a neurologist. The neurologist ordered an EMG and it was abnormal. An initial diagnosis was genetic neuropathy.

### **Who made the diagnosis?**

Ultimately a neurologist who was also a geneticist made a definitive diagnosis of Late Onset Sandhoff disease.

### **How long was your journey to diagnosis?**

Forty years.

### **Looking back, what advice would you give yourself?**

I would tell myself not to ever accept hearing "I don't know what's wrong with you," or "It's all in your head." You just can't give up.

### **What difference would an earlier diagnosis have made?**

An early diagnosis could have connected me sooner with people knowing what I was going through and that would have been a huge help. It could have saved me so much emotional pain—especially knowing that I wasn't alone.



# Know the signs of Late-Onset Sandhoff Disease

## You Could Make the Rare Dx

### First signs

Early symptoms of Late Onset Sandhoff can include clumsiness and muscle weakness in the legs. Once diagnosed, adults often reflect on their childhood and may notice that they experienced symptoms much earlier, such as not being athletic and/or speech difficulties or a stutter as a child or teenager.

The mental health symptoms may present first, which can lead to an especially long road to diagnosis. About 40% of affected adults experience mental health symptoms such as bipolar or psychotic episodes.

### Gradual loss of skills

Over time, adults with Late Onset Sandhoff slowly decline. Adults frequently require more mobility assistance, i.e., cane to walker to wheelchair. Many people experience speech and swallowing difficulties, but few require a feeding tube.

### Diagnostic pathway

Late Onset Sandhoff may be hard to diagnose. Some adults go five or more years before learning their true diagnosis. The disease may sometimes be misdiagnosed as Multiple Sclerosis or ALS.

### Other forms of Sandhoff

There are three forms of Sandhoff disease. Infantile Sandhoff most often manifests with symptoms appearing around six months of age. With Juvenile Sandhoff, symptoms appear after the first year of life, typically between the ages of two to five, but can occur anytime during childhood. With Late Onset Sandhoff, symptoms typically appear in adolescence or early adulthood, but they can also occur later in life.

### Risk profile

Anyone can be a carrier of Sandhoff. When both parents are carriers, each child has a 25% chance of having the disease. The carrier rate for the general population is low, approximately 1/600, and it is not yet clear whether Sandhoff disease is more common in any particular population, but it may have a higher carrier rate in several somewhat isolated populations.

