

# Living Life to the Fullest

## with a Rare Blood Disorder, Thanks to Innovative Therapies

**W**hen doctors see the same diseases day after day, diagnoses can happen quickly. But what happens when a condition is rare? For Barry Katsof, a Montreal businessman, getting answers was a two-year process that ended with a paroxysmal nocturnal hemoglobinuria diagnosis or PNH — a disease that affects only about 100 Canadians with a yearly incidence of 1 to 1.5 in a million.

In May 2001, he was experiencing dark urine in the morning. Doctors didn't know what it was. After seeing two different urologists, a kidney specialist, and then undergoing a number of tests, everything came back negative. "I was determined to get some answers," Katsof says. "I knew something wasn't right, so I kept pushing. I knew that having blood in your urine was not normal."

Finally, a hematologist was able to provide an accurate diagnosis of PNH, a life-threatening blood disease, by determining there was hemoglobin in his urine. This disease originates from bone marrow stem cells that acquire a mutation on the PIGA gene that causes red blood cells to be susceptible to attack by the immune system, thus destroying them faster than the body can replace them. It most often affects younger adults age 35 to 40, and it's difficult to diagnose because of the wide variety of non-specific symptoms, from fatigue and shortness of breath, to severe headaches and abdominal pain, to hemoglobin in the urine.

### Challenges in diagnosing rare blood disorders

In early 2003, few doctors were aware of PNH — and worse, there was very

little they could do to treat it. That was especially troubling because Katsof faced some serious fallout due to his condition, including chronic anemia and blood clots, a major cause of death among those with PNH. The average survival after diagnosis is about 10 years, though patients can live for decades with just minor symptoms.

PNH is a progressive disorder, so by 2007, Katsof was reliant on blood transfusions every two weeks. At that time, they were the best treatment available and the most they could do to curb the chronic anemia that comes with PNH. "I wasn't in good shape," he explains. "I could barely walk up a flight of stairs and anything I planned, like going out to dinner, was subject to how I was feeling and whether I could stay awake long enough."

### Living life to the fullest with PNH

In 2008, he started a new therapy that blocks the immune system from attacking and destroying red blood cells. The results of this treatment were encouraging. "I'm turning 70 in March and I now lead a full, normal life," says Katsof. "I cycle. I hike. I'm active. Life has reverted back to the way I was when I was 50 years old."

Atypical hemolytic uremic syndrome (aHUS) is another rare disease that is characterized by blood vessel cells being attacked by the immune system, through a similar mechanism to that of PNH. There are challenges when diagnosing this disease because of its various presentations and symptoms. This genetic disease is equally

life threatening, with more than 60 percent of aHUS patients dying, needing kidney dialysis, or having kidney damage within a year of diagnosis. It is also just as rare — affecting one in one million, split evenly between adults and children.

Katsof's journey demonstrates that there is hope for rare blood disorders like PNH and aHUS. He's keen to play his part in raising awareness by sharing his story and helping fellow PNH patients through the founding of the Canadian Association of PNH Patients, which provides support and assists with connecting them to doctors with specific expertise in PNH drug ther-



**Barry Katsof**  
President,  
Canadian Association  
of PNH Patients



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apies. With better medical treatment available, those with rare diseases can feel good about what lies ahead after a diagnosis. As Katsof points out, "The miracle of these new treatments is that they give people their lives back. Never give up hope!"

*Michele Sponagle*