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For Capt. John “Jack” Sautter, An Unexpected Diagnosis Becomes a Call to Action *After Discovering That He Had Polycystic Kidney Disease, a Marine Seeks a Better Future for Himself and Others*



Jack Sautter and daughter Katherine

There are many ways to describe John Sautter. First of all, call him Jack. Jack is a Captain in the U.S. Marines and a third generation veteran. He is a lawyer, working as a prosecutor in the Marine Corps. He holds a Ph.D. in political science. He is also a proud son, brother, husband, and father. Until a little over 7 years ago, Jack was also the picture of perfect health, or so he thought.

In one day, though, his life changed forever; he was diagnosed with polycystic kidney disease (PKD), a disease that has been traveling in his family for at least four generations. Rather than simply accept the diagnosis as an insurmountable obstacle, Jack approached this new challenge the same way he had approached every other one he'd faced: he searched

for ways to address it. One path he chose was to volunteer for an NIDDK-supported clinical trial of treatment options for people with PKD and, in doing so, try to build a better life not only for himself but for all people—both now and in the future—with the disease.

Diagnosis

On a crisp fall day in 2007, a then 29-year old Jack enjoyed a spirited rugby game with some friends from law school, followed by a backyard barbeque. He returned home, tired from the evening's activities, and tumbled into bed. When he woke the next morning, his foot was in so much pain that he could barely walk. "I thought I'd broken my foot," playing rugby the previous day, he says. He wasn't particularly worried, though, as he'd injured the same foot playing football years earlier. Still, he thought it best to have it checked out and headed off to the local hospital's emergency room.

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After a brief physical exam and evaluation, the pain in Jack's foot was diagnosed as gout, which surprised him. Gout is caused by the deposition of crystals

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of uric acid in the joints—usually in the extremities, commonly in the toes—which causes inflammation and sometimes quite severe pain. These crystals form as a consequence of elevated levels of uric acid in the blood. Usually, uric acid remains dissolved in the blood and is filtered and excreted by the kidneys. High levels of circulating uric acid are sometimes, but not always, an indication of underlying kidney problems.

As the emergency room staff asked questions about Jack’s medical history, he told them that several members of his family, including his father, had been confirmed to have PKD, and several other relatives were thought to have had it. “The doctors rolled in an ultrasound machine and held it up to my kidneys...and I could see the cysts, right there.” This unexpected news was traumatic and scary, and his initial reaction was one of “panic and fear.” He adds, “I did *not* go to the hospital thinking that I was going to be diagnosed with PKD.” In that instant, Jack went from a self-described “carefree, young, rugby-playing Marine officer who could do anything, accomplish anything” to someone whose “life was inexorably changed.”

There was another reason why this diagnosis was particularly unsettling for Jack. Cardiovascular disease is often seen in people who have longstanding PKD, and it had been just 3 years since his father had passed away after a heart attack. He was 56 years old when he died.

Polycystic Kidney Disease

PKD is a genetic disorder characterized by the growth of numerous fluid-filled cysts in the kidneys. There are two main forms of PKD. The most common is autosomal dominant PKD, which is the kind in Jack’s family. Symptoms usually develop between the ages of 30 and 40, but they can begin earlier, even

in childhood. In the United States, about 600,000¹ people were estimated to have PKD in 2000, and cystic disease is the fourth leading cause of kidney failure.

In most cases of autosomal dominant PKD (henceforth referred to simply as “PKD”), the slow progression of cyst growth can go unnoticed for many years. Many people with early-stage PKD have no symptoms, and their physical condition appears normal. The cysts, which can number in the thousands, can profoundly enlarge the kidneys while replacing much of their normal structure, resulting in reduced kidney function and potentially leading to kidney failure and a host of other health problems. Jack says that, when his father was in his 50s, his kidneys were estimated to be the size of small footballs. “You could actually see his kidneys bulging out of his sides,” he adds.

Many people with PKD experience a decline in their kidney function as the cysts grow, and about one-half of them progress to kidney failure and require dialysis to live. High blood pressure is another common health problem for people with PKD. In most people with PKD, high blood pressure appears by age 20 or 30; it can lead to serious cardiovascular complications such as heart attack or stroke, both of which contributed to Jack’s father’s death. Other complications of PKD include urinary tract infections, blood in the urine, and kidney stones.

Jack Sautter Takes Action

Fortunately for Jack, tests at the hospital revealed that his kidney function was normal, suggesting that his PKD had been detected before serious damage had occurred. After the shock of his diagnosis had passed, “I started doing what everybody does in the modern age: I jumped onto the Internet and started reading everything I could.” He scoured the websites

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of hospitals, news organizations, and even blogs in an effort to learn as much as he could about the disease.

In November of 2007, Jack found a set of slides on the website of Tufts University School of Medicine. The presentation was from a conference that the school had hosted on PKD; these particular slides talked about a clinical trial called HALT-PKD. He thought, “Wow, this is really fascinating!” At the time, Jack was attending law school in Vermont; Tufts is located in nearby Boston, Massachusetts. When he discovered that Tufts was one of the sites participating in the trial, he located contact information for the university’s HALT-PKD site and picked up the phone. Shortly after that conversation, he had enrolled in the study.

The HALT PKD Trial

Launched in 2002, the HALT-PKD trial enrolled two groups of volunteers based on their kidney function: those with relatively healthy kidneys were enrolled into “Study A,” and those with more advanced disease and diminished kidney function were enrolled into “Study B.” Jack entered “Study A,” which recruited 558 volunteers with early-stage PKD at seven medical centers around the country. This study tested whether drugs that target the renin-angiotensin system—an important regulator of blood pressure and fluid balance—could slow the progression of the disease.

In the HALT-PKD Study A, the volunteers were given oral medications aimed at lowering their blood pressure, which is expressed as a higher number “over” a lower number. Half were assigned to a group with the goal of achieving “standard” blood pressure, defined as between 120 to 130 over 70 to 80. The other half targeted a lower blood pressure, between 95 to 110 over 60 to 75; while lower than the “standard” target, this is still within the normal range. Within each

group, the participants were started on either a single drug or two drugs to reach their goal. (Those receiving a single drug received a placebo pill as their second “medication.”) Jack was randomly assigned to the low blood pressure group. “I was hoping to be assigned to this group,” he says. “I thought that perhaps I could benefit from the study by having lower blood pressure,” although at the time he signed up he had no way of knowing what the outcome of the trial might be.

Jack was an enthusiastic volunteer in the HALT-PKD study. Even a 7-month deployment to Helmand province in southern Afghanistan from October 2011 to May 2012 could not prevent him from continuing to participate.

Throughout the trial, Jack and his fellow study participants had regular check-ups, provided blood samples every 6 months, and underwent magnetic resonance imaging (MRI) of their kidneys to monitor cyst growth several times. Jack was an enthusiastic volunteer. Even a 7-month deployment to Helmand province in southern Afghanistan from October 2011 to May 2012 could not prevent him from continuing to participate. He asked his physician in the United States to forward his medical records to the unit’s battalion surgeon, who arranged for Jack to visit the local combat hospital. There, he was able to provide blood samples that were analyzed on site; the results were forwarded to the HALT-PKD investigators at Tufts. When Jack returned home, he continued his participation in HALT-PKD until the trial’s end in 2014.

Data collection in HALT-PKD ended in the late summer of 2014. The investigators spent several months analyzing the data, and the results of the trial were

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announced at the annual scientific meeting of the American Society of Nephrology in November 2014 (see below for more information). Jack is “really looking forward” to learning the results, because they may benefit not only him personally, but also his family members and the larger numbers of people with PKD.

PKD and the Sautter Family: Past, Present, and Future

PKD has a long history in the Sautter family. Jack’s great-grandmother, a member of the first generation of the family to be born in this country, is thought to have had PKD, although she was never formally diagnosed. His grandfather, an Army pilot in World War II, had PKD. His father, a 20-year Army veteran, had it. Jack is acutely aware of the toll exacted by advanced PKD, as he had helped care for his father during the last years of his life. Speaking of him, Jack says, “A lot of things that slowed him down and made life more difficult for him were related to PKD.” These experiences made Jack acutely aware that PKD affects not only the people with the disease themselves, but their families and caregivers as well.

After a tour in Okinawa, Japan and Hawaii, Jack is currently stationed at Camp Pendleton in California, where he lives with his wife and 2-year-old daughter. When he reflects on his participation in the HALT-PKD trial, he knows that, even if the findings from the trial do

not directly help him in the near term, his participation in this research might help other people with PKD in the future. While he says that one motivation for entering the trial was that it might benefit him, he is quick to add, “Like in all things, our decisions are not as simple as just one motive; there are lots of motives.... It wouldn’t shock me if my daughter has PKD.” And, while he hopes that she stays healthy, if she is someday diagnosed, “hopefully, she will benefit from this research.” He adds, “That was definitely on my mind when I was trying to become a part of HALT-PKD and continuing to do the study.”

The NIDDK and the PKD Foundation sponsored the HALT-PKD trials, which consisted of two treatment trials for autosomal dominant polycystic kidney disease. HALT-PKD was the largest and longest study of treatments for this condition. The results were initially reported at the annual scientific meeting of the American Society of Nephrology in November 2014, and were published as this document was going to press. More information about the study outcomes is at www.nih.gov/news/health/nov2014/niddk-17.htm

¹ Grantham JJ, Nair V, Winklhofer F. Cystic diseases of the kidney. In: Brenner BM, ed. *Brenner & Rector’s The Kidney*. Vol. 2. 6th ed. Philadelphia: WB Saunders Company; 2000: 1699-1730.