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PatientVoice

Changing Health Disparities in Autosomal Dominant Polycystic Kidney Disease (ADPKD)

Suzanne F. Ruff

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Mooresville, North Carolina

Correspondence: Ms. Suzanne F. Ruff, Mooresville, NC. Email: ruffsuzanne@gmail.com

The data in the study "Health Disparities in Autosomal Dominant Polycystic Kidney Disease (ADPKD) in the United States," published in this issue of *CJASN* (1), encourage changes. Recognizing the disparities in autosomal dominant polycystic kidney disease (ADPKD) can be building blocks.

The hereditary part of ADPKD is a major factor of this disease, especially in my family, where we can trace the disease to the 1940s when my grandmother's kidneys failed because of ADPKD. There was nothing the doctors could do to save her. At that time during World War II in Europe, Dr. William Kolff, frustrated with kidney failure in one of his young patients, was inventing an artificial kidney: the dialysis machine.

Five of my grandmother's six children inherited the gene that causes ADPKD, including my mother, her youngest child. Those six siblings in the 1960s and 1970s, before computers and cell phones and with almost no resources, united to educate themselves, with deep respect for each other's differences, choices, and limitations. Disparities occur within families: different geographical locations, education levels, and incomes. Their examples of living, coping, and yes, even dying with this disease are praiseworthy.

Within the ADPKD community, however, are patients with a mutation of the gene and absolutely no family history who are bewildered and shocked to be diagnosed with ADPKD. Then, there are others, maybe someone adopted or someone with one absent parent who carried the gene, who knew nothing about the hereditary ADPKD in their genetic makeup.

Everyone handles the diagnosis differently, including the person who gives the diagnosis. Baffled, one patient is told there are a lot of cysts on the kidney. Many in the health care field do not know what ADPKD is! Often, no one suggests seeing a nephrologist. Once, a nephrologist commented that he remembers reading only "one paragraph" in medical school about polycystic kidney disease. My sister suggested, "I'll teach you more. Let's be a team!" I also heard a nephrologist comment at a conference that "ADPKD patients don't suffer from pain." A lively discussion followed!

Disparities can change when health care providers learn exactly what ADPKD is, along with ways to delay and prepare for kidney failure. Being a coinvestigator for a recently completed 5-year research study called "PrepareNow: Putting Patients at the Center of Kidney Care" (2) has been a gratifying way to enable patients to learn more about transplantation and have an active role in their choices to treat kidney failure. My mother always said losing "control" was the hardest part of kidney failure. To me, that is the quintessential example of any patient's frustration.

ADPKD can be accidentally diagnosed through a mishap (one uncle was injured during a baseball game and received his ADPKD diagnosis; one cousin was injured and diagnosed while playing hockey). It can be a terrifying diagnosis for a patient with or without a family history.

Once, a twentysomething-year-old was diagnosed with ADPKD because of off-the-chart high BP and was told, "Take this blood pressure medicine, ADPKD won't happen for years and there's nothing you can do about it." A coworker of mine was diagnosed with ADPKD on her lunch hour. She was told, "It won't affect you until you're old. Come back when you can't eat your dinner."

We tell them not to believe in what my sister's nephrologist calls the "doc-in-the box" or dire information websites. Taking control and accountability are hard to do when you are floundering. Understanding the denial and depression that often comes with ADPKD (and any CKD) is vital, too, along with respect, compassion, and patience for the different ways people handle their diagnoses.

The psychologic aspects of a genetic disease are often unrecognized. The diagnosis of ADPKD, or the possible diagnosis of ADPKD, opens another can of disparity worms. It epitomizes my favorite expression: "When a patient is diagnosed with kidney disease, the family has kidney disease."

However, we have come a long way. In the 1960s when my mother learned that ADPKD was genetic, she was told, "There's nothing anyone can do about it until you're sick, but you shouldn't have had children, nor should your mother have had any." Mom, a feisty mother of three, almost reached across the table and grabbed the geneticist by his lapels; she replied, "Who are you to tell me my first years of life weren't worth living?"

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My advocacy is a result of frustration when people ask, "PK what?" ADPKD is considered a "rare" disease by those outside the community of people living with it. However, over 20 of my family members have or had ADPKD, with ten deaths, including my mother and sister. Today, my other sister and three cousins battle the disease. ADPKD is now into the fourth generation.

I write about what ADPKD does to families to help nephrologists and the medical community better understand and unite with their patients to fight the disease and most of all, to find a cure for ADPKD.

Disclosures

S.F. Ruff serves on the Executive Board of Directors at the American Association of Kidney Patients. She is the author of the nonfiction book *The Reluctant Donor*. She serves as a coinvestigator on the PCORI research study STEPS and is a family member of Kidney PAC at Otsuka.

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Author Contributions

S.F. Ruff wrote the original draft.

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