

What a Genetic Kidney Disease Does to Families:

An Invited Editorial by Suzanne Ruff

"An elephant in the room" is an idiom defined in the *Cambridge Dictionary* as "an obvious problem or difficult situation that people do not want to talk about" (Cambridge University Press, 2006). Many families with a genetic disease describe this attitude among themselves. Anguish, fear, and misunderstandings often result, causing more stress to a family touched by disease. Nephrology social workers, as well as all medical personnel, can benefit from understanding these issues when dealing with families like mine. My family battles a genetic disease called polycystic kidney disease (PKD).

PKD is one of the most common life-threatening genetic diseases, and the fourth leading cause of kidney failure (PKD Foundation, n.d. a). About 1 in 500 people have PKD (PKD Foundation, n.d. b). Individuals with PKD have cysts in their kidneys that can make the kidney very large, which causes kidney failure (NIDDK, 2015).

There is no cure for PKD. Dialysis and transplantation are the only treatments. My family suffers from the most common form of the disease, autosomal dominant polycystic kidney disease (ADPKD). If a parent carries the gene for ADPKD, each of their children has a 50% chance of inheriting it (PKD Foundation, n.d. b). There is also a recessive form of the disease, autosomal recessive polycystic kidney disease (ARPKD). It is rarer than ADPKD, but causes devastation for patients and families as well.

Those are the facts of PKD. Facts are cold and hard. Stories of the people behind those facts can both break your heart and inspire you. No one likes to hear the word "disease." Adding the word "genetic" has an even more devastating impact on individuals and families.

Sometimes a family doesn't realize how a genetic illness impacts so many aspects of their lives, because each member reacts differently. That's where misunderstandings happen. Nephrology social workers can help patients and families cope with this diagnosis.

Some members of a family with PKD come out fighting against the disease—fists raised, ready to do battle, and announcing it to the world. They educate themselves about the disease, and loudly shout its horrors from the rooftops. "Great Aunt Sally must've died from this genetic disease," they exclaim, adding, "Didn't Uncle Harry have kidney issues too?" Sometimes they don't get tested because of the never-ending worry about getting insurance. They face the disease directly and talk about it (sometimes incessantly or even obnoxiously).

Other family members deal with PKD more privately. They need time to digest the news, and they don't want to talk about it publicly, or they don't want to talk about it at all. They don't want others to know they have the disease. Nor do they want to know how "Uncle Harry or Great Aunt Sally died." They might think, "She's dead; what difference does it make?" It's not that they don't care; it's just too painful.

Encouragingly, healthcare and kidney disease treatment have entered a new phase that emphasizes *patient-centered care*, and patient and family member engagement (Narva, Norton, & Boulware, 2015). With the help of the Patient-Centered Outcomes Research Institute, there is a growing emphasis on including patients and their family members in all aspects of research. As a PKD family member, I share our story in the hopes of informing future social work research and practice to help patients with genetic kidney diseases.

My family has over 20 members with the disease. There is no right or wrong way to handle a genetic disease. With families like mine, where nine deaths have occurred (from the 1940s to 2013), some of the questions asked by medical professionals are painful. Patients consider their care team to be knowledgeable, and the one question the patient often asks is: Does the disease skip a generation? No, it does not. There is a 50% chance of inheriting the disease.

Questions often asked of a PKD patient and questions most asked by PKD patients are "Have your children been tested?" and "Should I have my children tested?" Oh, what questions! Debates rage among families about this subject. Individuals in families with PKD have different opinions including: "I can't bear knowing," "I can't bear not knowing," "We can plan our life accordingly," or "They deserve to be children without worries."

In the "old" days (early 1970s), my parents didn't feel it would be wise for us to know if we had a "preexisting condition." We were tested by archaic standards compared to today's medical care. Our parents kept the results of our tests to themselves. They didn't think we could handle the truth as teenagers. That is the dilemma of testing and telling children. Can they handle knowing they have a disease? Even with the changes in today's healthcare laws regarding preexisting conditions, many families still hesitate to get tested or inform their children about the results. What can a person do differently if they have the disease? Eat right? Maintain good blood pressure? Everyone should do these things, with or without polycystic kidney disease.

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Today, with the increasing numbers of living donors, many PKD families have started saying, “you’re either the recipient or the donor,” i.e., if you didn’t inherit the gene for PKD, and a sibling or a child has the disease, you have been “chosen” to be the living donor when the time comes for transplant. This adds another tremendous burden to families, and the psychological aspects of this expectation are significant. There are siblings or children who want no part of being a living donor for various reasons. There are PKD patients who will not accept, and do not want, a family member to be their living donor. It’s complicated, and the reasons are as numerous as the cysts that multiply and cause PKD kidneys to fail.

“Did you have a happy childhood?” This question was asked of a woman about to start dialysis because of PKD. When the woman was 12 years old, her mother died of PKD. Many have happy lives, but that “elephant in the room” is always present, even on the happy days. My mother was the youngest of six children; five of them inherited the disease, and their own mother was ill with PKD for most of their lives.

The question of having children comes up often in a PKD family. People will ask “Do you know you shouldn’t have children?” or “Why did you have children?” My mother and her siblings had already started their families before they even knew or heard the word “genetic.” Mom’s kidneys began to fail when she was 50 years old. In the early 1970s, when a genetic counselor advised my sisters and me not to have children, my mother bristled and said, “Who is to say my first 50 years of life weren’t worth living?”

Then, there was the guilt that crushed her — completely unwarranted guilt — for passing the disease to my sister. Mom died before knowing another of her daughters had PKD. If PKD hadn’t killed her, that probably would have. Ten years ago, my niece, who has a 50% chance of inheriting PKD, broke down in tears after her engagement, stating she “really wants to have children.” And, she did. “I am encouraged by the progress that is being made in finding a cure, and hope there will be a cure when and if my children inherit the disease,” she explained. She works relentlessly to raise money for PKD research.

PKD can also be caused by a gene mutation, with no past family history of PKD. One woman, another dedicated fundraiser for a cure for PKD, explains this bluntly. Karyn Waxman, 60, was diagnosed 16 years ago with PKD. With no apparent family history of the disease, a mutated gene resulted in the diagnosis. She states, “I’m okay with it . . . PKD starts somewhere as a mutation, but sadly ends up as ‘the gift that keeps on giving’ in many cases. However, the most difficult day of my life was the day that I sat both of my daughters down for a very frank discussion with them about the ramifications of this genetically inherited life-threatening disease. Knowing that I could have inadvertently passed it along to one or both of them was, and continues to be, a devastating thought. Now, when I look into the eyes of our two precious baby granddaughters, I hold back tears

and pray that their mother dodged what I consider to be my ‘PKD bullet.’ It’s awful beyond words when you bear the sole responsibility for possibly being the cause of pain for those you love and cherish the most in the world.”

A bullet! My mother could not have said it better.

An elephant, a bullet, whatever you call it — the guilt becomes a psychological burden that shapes everyone within these PKD families, whether they inherited the disease or not. Patience, tolerance, and wisdom are needed when dealing with a family with a genetic disease. Social workers and other medical professionals need to understand how PKD affects family dynamics, and bring comfort to patients and their families. You are an essential, important, and exceptional part of a kidney patient’s life. My family thanks you!

AUTHOR NOTE

The author is a member of the National Kidney Foundation’s Living Donor Council Executive Committee, a member of the Polycystic Kidney Disease Foundation, a board member of the American Association of Kidney Patients, the author of The Reluctant Donor, and was a living donor athlete at the Transplant Games.

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