As someone diagnosed with autosomal dominant polycystic kidney disease (ADPKD) almost 20 years ago, I am grateful for the opportunity to provide my perspective on the CJASN article “Presymptomatic screening for intracranial aneurysms with autosomal dominant polycystic kidney disease” (1). Intracranial aneurysm is an extrarenal manifestation of ADPKD. Moreover, because intracranial aneurysm rupture is associated with a high level of morbidity and mortality, it remains an important element of preventive ADPKD research. Therefore, this study by Torres et al. (1) is a follow-up from their previous work in 1989–2009 (2). This study reports on patients with new cases of intracranial aneurysms diagnosed between 2009 and 2017 and substantially extends the follow-up from the previous study.

After viewing the medical records of 3010 patients with ADPKD for almost 30 years, Torres et al. (1) identified 812 patients with ADPKD who went through presymptomatic magnetic resonance angiography screening. The patients with ADPKD who were selected either had a family history of intracranial aneurysms or subarachnoid hemorrhage before elective surgery or kidney transplantation or had a high-risk occupation. The magnetic resonance angiography screening resulted in identifying 75 (9%) patients who had unruptured aneurysms of ≥2 mm. Hypertension and smoking were more common in patients with aneurysms. In addition, patients with intracranial aneurysms had more advanced CKD. The results in this study mirror previous work. The prevalence of intracranial aneurysms is four times higher in patients with ADPKD versus the general population (8%–12% versus 2%–3%, respectively).

Because of the limitations of the design of this study, the authors were unable to draw firm conclusions on the effect of aneurysm screening. For example, the natural history of intracranial aneurysms in patients with ADPKD versus the general population remains unknown. This lack of knowledge makes it difficult to understand how interventions may differ in patients with ADPKD versus the general population. Consequently, the authors recommend targeted prescreening in the aforementioned population of patients with ADPKD. I am in agreement with their recommendation.

I found the expanded observations and the modifiable risk factors for intracranial aneurysms to pique my interest. I learned that hypertension, smoking, and alcohol intake are modifiable risk factors for intracranial aneurysms. This captured my attention because of my desire to shift kidney disease interventions upstream rather than waiting for downstream complications to occur. Moreover, this article also provided me an opportunity to reflect on my own patient journey and lessons that I learned. I will share how a family narrative can influence behavior and the opportunity for the ADPKD global community to now shape behavior through a new patient narrative.

My journey started in the 1970s when I witnessed my mother suffering from ADPKD. At that time, I was a teenager transitioning to a young adult. The family narrative was that there was nothing that you could do to treat ADPKD, and for the most part, that narrative was accurate. This family narrative started with my grandfather in the 1950s. My mom shared stories of her dad being admitted to the hospital in the late 1950s, because he was not feeling well. Unknown to my grandfather and his family, he was in the initial stages of kidney failure and dying of uremic poisoning. Although he had ADPKD, he was never diagnosed. He was a heavy smoker, and I am sure that smoking accelerated the decline of his kidney function.

As my mom’s ADPKD progressed, she suffered nausea, fatigue, pain, and depression. It was never clear to our family what her health outcome would be. There was this black cloud of uncertainty that existed in our family, and it permeated our life. My mother’s health was never discussed, because it was a taboo subject. It was taboo, because there was nothing you could do but wait as ADPKD progressed to kidney failure.

In 1981, my mother started hemodialysis. Although dialysis sustained her life, her suffering only intensified. I have deep memories of her coming home from dialysis tired and nauseous from her treatment. I have other memories of her arm bloodied and bruised after a rough dialysis treatment. After 4.5 years on dialysis, my mother died in 1986 at the age of 52 years old. After seeing her suffering, I lived in constant fear that I would experience the same fate.

While I was a young man living in the 1980s and 1990s, I placed the possibility of ADPKD in the back of my mind. Although I never smoked, I did drink my fair share of beer and caffeinated soft drinks, and I ate a lot of fast food. Although my primary physician was
aware of my ADPKD family history, he said that my slightly elevated BP was nothing to be concerned about and that my kidney function was fine.

Shortly after receiving a clean bill of health from my physician, I began to experience persistent back pain. At that time, I had been married for 5 years, and I had a 3-year-old daughter and 9-month-old son. I asked my physician to perform an ultrasound to determine if I had ADPKD. After the test was completed, my doctor diagnosed me with ADPKD and informed me that I would be in kidney failure in 3–5 years. Because my physician had provided me contradictory information on my kidney function, I declined his nephrology referral and sought my own nephrologist. Through the help of friend, I found a nephrologist who had high expectations for me, and I received a preemptive kidney transplant. This moment of patient activation created a family narrative that was empowering and hopeful.

Today, there exists an opportunity to activate the global community of patients with ADPKD. The PKD Outcomes Consortium was formed between the PKD Foundation and the Critical Path Institute. The consortium has successfully qualified total kidney volume as a prognostic biomarker with both the US Food and Drug Administration and the European Medicines Agency. This biomarker has resulted in one upstream treatment with the potential of more to follow. This powerful message of hope needs to be understood by the patient community while specifying the roles and responsibilities of patients with ADPKD to accelerate kidney health.

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