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Letter to the Editor

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An Alport Syndrome Journey: From Powerless to Empowered – A Patient Perspective

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Abstract

As an Alport syndrome patient, caregiver, and executive director of Alport Syndrome Foundation, I am aware of the frequently challenging road in seeking an accurate diagnosis. Our journeys are scattered with misdiagnosis, missed opportunities for accurate diagnosis, counterproductive medications, and overwhelming guilt when our children are diagnosed. We understand that most of our healthcare providers know very little about our disease. Typically, it is incumbent upon us to become empowered through education and connection with our patient community to be sure that our physical and emotional health is well managed.

Dear Editor,

Misperceptions persist that an accurate Alport syndrome diagnosis is not critical to care for our orphan disease. As patients, we see this differently. Accurate diagnosis opens the door to important options such as understanding recommended treatment guidelines from experts, identifying other family members at risk, awareness of potential complications for monitoring or treatment, and opportunities to participate in research. Genetic testing is much more accessible and a key component in understanding our rare disease. Empowered patients can help themselves and their providers, leading to better outcomes.

Misdiagnosed

For the majority of my 56 years, I was misdiagnosed with the wrong kidney disease. Most of that time, I did not ask questions, did not do my own fact finding, and was passive about my health. In sum, it did not go well. When I became a more educated patient, willing to self-advocate and actively participate in my treatment and care, many aspects of my life improved.

As the executive director of Alport Syndrome Foundation (ASF) since 2019, an organization led by and dedicated to patients and families, I often hear stories similar to my own. Our journeys are scattered with misdiagnosis, missed opportunities for accurate diagnosis, counterproductive medications, and overwhelming guilt when our children are diagnosed. Although an Alport syndrome diagnosis can be difficult to accept, it can also provide an unexpected sense of relief to finally know the true cause of one's symptoms and the ability to mindfully carve a path forward for what lies ahead.

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Misinformed

In dealing with the medical system, it becomes abundantly clear that Alport syndrome is rare. Most clinicians I meet admit they have never heard of it or remember a question from a medical board exam but cannot quite put together what it was. An Alport patient I know who is currently studying to be a nephrologist noted that our disease was given one paragraph in his medical school textbook, and the information was outdated.

In October 2021, on one of the many occasions that my then 19-year-old son was in the emergency room facing end-stage kidney failure, an internal medicine specialist entered the treatment room and proudly shared his knowledge about Alport syndrome. He addressed my son, "I bet you don't have any sense of smell. I know that's how many Alport syndrome patients are diagnosed." My exhausted son responded, "Respectfully, doctor, you are confidently mistaken." When I tried clarifying that perhaps he was thinking about hearing loss, which is associated with our disease, the doctor argued with me and walked out.

My Story

It was the 1970s when my kidney disease journey began. I remember being in early elementary school and shopping for clothes with my mother at a department store. The sales assistant undiplomatically pointed out that we were in the wrong section. I could no longer browse in the "slim" section. I was now relegated to options for "larger girls," which were less appealing to me than the styles I wanted to wear at the time. I was upset. My mother explained the culprit was a full year of high-level prednisone to treat my kidney disease. All I knew was that I felt crummy, the shape of my face and body had completely changed, and I did not understand the talk about kidneys I kept hearing at doctor visits. In more adult terms, beyond me at that point, I had been diagnosed with IgA nephropathy or "Berger's disease" by renal biopsy.

Two years into steroid therapy, with no positive change in my laboratory results, the doctor unceremoniously halted the medication. No more drug therapy, and no more answers. I continued to spill urine that looked like Coca-Cola when I had viral infections.

Like many families facing chronic kidney disease, we drove hours to see a pediatric nephrologist when I was young. This meant time off work for my mother, a widowed single parent. We would do the 8-hour roundtrip drive from our small Midwest town to a city with a university hospital. More specialists, more visits, but never much more guidance.

I went off to college and lived my life, never dedicating time or resources to visit a nephrologist. I did not understand why I should prioritize expensive and uninformative medical appointments.

Misgivings

In my early 20s, I moved to a large urban area post-college. I decided to be brave and visit a teaching hospital. I was working with children, which got me thinking about whether or not this "kidney thing" was going to present an obstacle to motherhood. All I knew were the words "IgA nephropathy" and "Berger's disease." My blood and urine test results were shared with the nephrologist, who did not speak to me at all. While I squirmed self-consciously on the noisy white paper covering the examination table, the doctor directed his remarks to the students in the room. He noted it was highly likely that I would be in renal failure before the age of 30, and having children would be a real question mark. I could barely keep my emotions in check, let alone ask questions. There was no follow-up suggested. I was too scared to visit a nephrologist again for many years. I vowed to live as healthy a life as possible, motivated to prove that particular doctor wrong. I took up distance running and exercise classes, thinking this would somehow make up for the silent mystery going on in my body. For more than a decade, my approach to kidney disease was serious denial.

As that kidney patient in my 20s, I never found my voice. I did not feel educated enough about my disease to ask questions. This was years before conducting research online in the privacy of my home was possible. I never saw the same doctor twice and was shocked to be denied insurance at the non-profit organizations where I worked because my health records indicated kidney disease.

Misguided

By my mid-30s, I thought I had tricked fate. I gave birth to two beautiful sons 2 years apart. My second pregnancy was troublesome as I experienced pre-eclampsia and had challenges recovering after childbirth, but both my sons seemed healthy when they were born. When my second son began spilling blood in his diaper as

an infant, my heart sank. How could this be? Doctors along the way had informed me that my kidney disease was not genetic.

We were referred to a pediatric nephrologist who told me and my husband that our son must have "familial nephritis" and that it was likely benign. We should not worry unless he started spilling blood in his urine when he did not have an infection. We were told to check in once a year for renal laboratories to be on the safe side. We did what we were told.

When our son turned four and was frequently ill, his gross hematuria was evident and concerning to us. I met with the nephrologist assigned to us at the hospital and asked if biopsy or other diagnostic test could help us better understand what was going on in his kidneys. The night before a scheduled biopsy, a different nephrologist phoned my home and said she was scheduled to be the doctor on call the next day. She did not agree with the colleague that ordered the procedure, and refused to perform it. Her reason was "Your son has what you have. Why put him through a biopsy?" She was convincing. I did not push back.

Mystery Solved

Seven years later, the day I was stunned by a doctor's assessment that my now eleven-year-old son needed hearing aids, I went home and searched key phrases on the internet: "kidney disease, boy, age 11, hearing loss." There it was: Alport syndrome. Each article fit like a glove. I knew instantly, and with deep fear, that he was facing this rare kidney disease. I read dozens of published articles, doing my best to understand all the scientific terms that were new to me. If we had been accurately diagnosed, I could have been monitoring his hearing. My son's year-long struggle at school and withdrawal from his typical social behavior held new context.

By this time, as a more experienced parent, the courage to trust my own instincts had grown. I contacted the nephrology clinic and shared this new development and my concerns, requesting an appointment. A nurse called back stating that the next available appointment was 4 months out. In the meantime, she instructed, if my son experienced fluid retention, nausea, or unexplained itching, I should call back and perhaps they could fit us in. To be sure I understood correctly, I asked, "So I should only call if he's experiencing symptoms of renal failure?" The answer was "That's correct." The ear, nose and throat specialist who had diagnosed the hearing loss was equally unhelpful. "That disease is so rare. I'm sure he doesn't have it. You should avoid diagnosing your child through internet articles."

Moments of desperation can lead to transformation, panic to proactive approach. I began considering hospitals and doctors in other cities, reviewing reputations and biopsy infection rates. Through research and personal contacts, I discovered a new doctor had recently been hired at a different local hospital and was certified in both adult and pediatric nephrology. To me, a nephrological unicorn! I called and used the words, "Alport syndrome," which this time secured an appointment in less than a week.

Within 2 months, my youngest son was accurately diagnosed by biopsy. It seemed clear that he must have inherited the disease from me. With the X-linked form of Alport syndrome, I had a 50% chance of passing my genetic mutation to each of my children. Through genetic testing, we confirmed my older son was not affected.

The nephrologist encouraged us to join a patient registry and to contact a patient organization, noting we would need support. She was right. I am so grateful that she continues to encourage her newly diagnosed Alport patients and caregivers to get in touch with ASF.

It was painful to watch my son adjust to middle school while also dealing with hearing aids and new medications. His confidence was erased. With ACE inhibitor side effects causing him to pass out frequently, he gave up organized sports and often had to sit out during physical education classes after embarrassing experiences with fainting at school. His teammates left him behind as he could no longer do what they were doing. He carried around clunky equipment from class to class that plugged into each teacher's microphone to combat school background noise. Fear of his hearing aids getting wet or malfunctioning was constant.

Motivation

I found there is true power in educating yourself about your kidney disease and/or your child's disease, particularly with rare conditions. After our accurate diagnosis in 2014, education and community made a significant difference in our lives. With Alport syndrome, there is a great deal of new content to comprehend: medications, side effects, laboratory values, genetic variants, disease-specific terms, potential complications, recommended tests, myriad specialists, hearing devices, and anxiety about all of them.

The importance of understanding that you are not alone may sound cliché, but when it is happening to you

and your family, and feelings of isolation and fear are all-consuming, the voice of someone else that speaks your new language is the most comforting sound in the world. Strangers or not, you are immediately kin. I have often been on the phone with other patients or caregivers who are emotional because it is the first time they have spoken to someone else that shares our rare disease.

When my youngest son was twelve, about a year into his diagnosis, we attended our first meeting organized by ASF. We met nephrologists with expertise treating our disease, researchers advancing studies, and other families like ours trying to learn everything they could about living with this rare condition. Both our sons spent 2 days with other teens and their siblings affected by Alport syndrome. At the end of the first day, my younger son said to us, "I had fun today. Thanks for bringing me here. I don't feel so alone anymore." It was music to my ears and the beginning of a major, positive shift for our family.

At the patient meeting, my son transitioned from hiding his disease at all costs, to becoming an active and vocal patient advocate. Listening to researchers explain their work, he realized if patients are not open about their disease, there would be no way for scientists to find new medications or a cure. On the drive home, he and his brother started hatching a plan to raise funds for research. Moreover, they pulled off that plan and brought new awareness and funds to ASF. Even more valuable was that our family became open with each other and the outside world about living with Alport syndrome.

Mission

My youngest son and I began attending legislative advocacy events for both kidney patients and rare disease families. For the past 7 years, we have learned, listened, spoken up, shared our stories, and advocated to members of congress for legislation that makes a difference for families like ours. It gives us hope and a sense of purpose, and shaped my son's attitude and goals for the future.

When the need for dialysis and transplant arrived quickly at age nineteen, my son had a network of people to support him through the physical and emotional challenges he faced. As a parent having to make critical decisions during an emotionally difficult time, I also had a helpful group of parents through ASF as a support system. They provided practical guidance, empathetic conversations from those who had "been through it," and so much more. My older son, who became his younger brother's living donor at age twenty-one, had also been educated about our disease over time. He had been emotionally and physically preparing for this possibility for years. He was on a mission to be the one to save his brother, and he did.

Not everyone needs or wants to meet with legislators or become so public about their disease in order to take control of their care. It is just one avenue of participation and empowerment. There are so many others. It is my personal and professional experience that patients and families that get involved with ASF become more educated about their disease, leading to better outcomes. They can participate in research at a variety of levels to advance understanding of Alport syndrome and have their experiences documented, including enrolling in ASF's patient registry (https://alportsyndrome.org) and/or natural history study (https://alportsyndrome.org/for-patients/neptune-natural-history-study/). They can learn from and find inspiration in each other, and use patient-focused educational resources to take control of their disease management.

Misperceptions Persist

With no FDA-approved treatment for our disease at this time, misperceptions persist that an Alport syndrome diagnosis is not helpful to patients. From a patient point of view, there are many reasons for clinicians to help them receive an accurate diagnosis. It is a key that unlocks the door to empowering options such as:

- Understanding recommended treatment options;
- Identifying other family members that may be at risk;
- Awareness of potential complications to be monitored and/or treated such as hearing loss and eye abnormalities;
- Opportunities to participate in research and clinical trials; and
- Options to connect with other patients and families for support.

Genetic test by blood, saliva, or cheek swab has become a valuable diagnostic tool for our community. It is increasingly more accessible financially. A genetic testing panel for Alport syndrome and other rare kidney diseases can be secured for $350 or less in the USA. Some testing programs also offer options for genetic counseling at no cost to review and interpret the results. Suggested guidelines for recommending genetic testing can be found in the 2022 Kidney International article [1], genetics in chronic kidney disease: conclusions from a conference titled Kidney Disease: Improving Global Outcomes (KDIGO) Controversies, referenced at the end of this article.

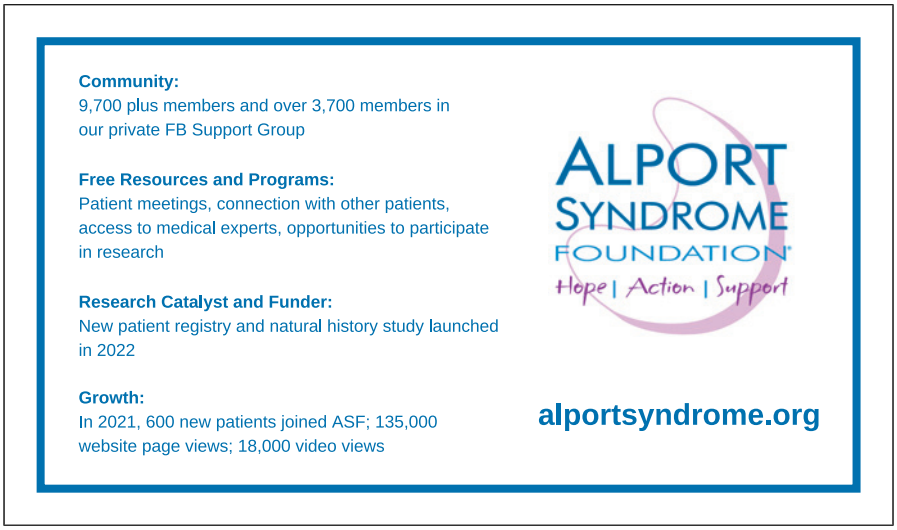

Fig. 1. A non-profit organization led by and dedicated to the Alport syndrome community of patients and families.

Patients have the right to understand if there is an underlying genetic cause of their CKD. If a mutation in a *COL4a3*, *COL4a4*, or *COL4a5* gene is identified, please encourage patients to contact ASF. We will welcome them with open hearts, extensive educational resources, and opportunities to get involved and/or connected in ways that work best for them. Empowered patients help themselves and their care team. I think we can all see that as a win-win.

In closing, I want to honor my patient advocate son, who is so disheartened when he learns that a medical provider has informed someone that they will likely never meet another Alport syndrome patient outside of their own family. We hear this too often, and it does not need to be true. It only makes people feel more alone. Please know there is a large and growing community of patients working together in the USA and internationally to advance research and treatment options, and to provide a network of support. We are here and empowered. Contact info@alportsyndrome.org. See Fig. 1 for more information about the efforts of ASF.

Conflict of Interest Statement

I am employed by Alport Syndrome Foundation to serve in the capacity as executive director.

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Author Contributions

Lisa Bonebrake is the sole author and contributor to this manuscript.

Reference

1 Köttgen A, Cornec-Le Gall E, Halbritter J, G Gharavi A. Genetics in chronic kidney disease: conclusions from a conference titled kidney disease: improving global outcomes (KDIGO) controversies. Kidney Int. 2022 June;101(6): 1126–41.