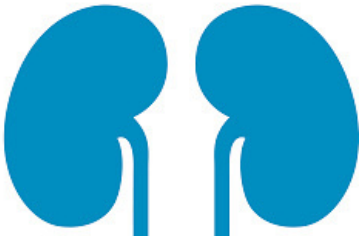
Pediatric Alport Insight Project

ALPORT SYNDROME FOUNDATION®

Hope | Action | Support

www.alportsyndrome.org

About the project



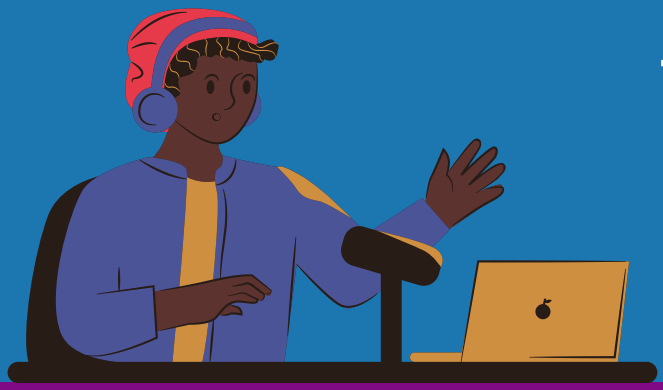
During a 2019 interview with Alport Syndrome Foundation Executive Director Lisa Bonebrake about his experience treating young patients, pediatric nephrologist Dr. Bradley Warady noted:

"*The psychosocial issues of pediatric Alport syndrome patients should be seen as a key aspect of the disorder and attention to these issues should be as proactively addressed as prescribed medications. These issues are as important, if not more important, than patients taking their ACE or ARBs.*"

Dr. Warady noted chronic kidney disease can compromise a teen's natural desire to develop independence. This in turn causes anxiety about their future, feelings of isolation, and possible depression.

Dr. Warady suggested that pediatric nephrologists welcome and benefit from pediatric patients' insights and experiences. Based on this perspective, Alport Syndrome Foundation took a proactive role in inviting 25 young Alport patients ages 13-22 to participate in the "Pediatric Alport Patient Insight Project." Beginning shortly before the pandemic, over the course of 2020 and 2021, these patients completed questionnaires, met with ASF Staff and other participants virtually, and shared their experiences individually and in small groups. This document presents a summary of captured insights.

Contents



The Alport Patients

Ages 13-22

15 Females & 10 Males

60% have family members with confirmed diagnosis of Alport syndrome

2 post-transplant patients, and 1 currently on dialysis awaiting transplant

52% have hearing loss related to Alport syndrome and wear hearing aids - 24% have eye abnormalties related to Alport syndrome

Frequency of Nephrology Visits:

40% - 1 X annually
24% - 2 or 3 X annually
36% - 4, 5 or more X annually

Diagnosis:

32%
genetic test

32%
renal
biopsy

24%
not sure

12%
skin biopsy

84% of participants report a parent was in the room for the entire visit during all pediatric appointments

Relationship with Nephrologist

survey results

96% feel comfortable enough to speak with a nephrologist without a parent or guardian in the room

Topics that a nephrologist has asked you about during a check up:

88% exercise/sports
84% school progress
76% diet /nutrition
60% hearing
56% feeling tired
29% depression

Relationship with Pediatric Nephrologist interviews

*He says stuff and **I listen**. - age 16*

*My relationship with my nephrologist is **very businesslike**. - age 17*

*My nephrologist would probably say I talk more than she does. I go in prepared with thorough questions or documentation of how I'm feeling. **We have a dialogue.** I couldn't ask for a better nephrologist. - age 20*

The doctor says the same thing each time.
***It's typically a 5-minute appointment.** - age 17*

***It's more my mom has a conversation with the nephrologist** and sometimes she asks me questions too. - age 16*

***We talk about the results of my lab tests** and what we can do better.*
Often very short appointments. - age 15

*My nephrologist did a really job good job of explaining things well, even drawing out diagrams on a piece of paper, so that **I eventually I became comfortable enough to lead my own care**. - age 18*

*My old nephrologist retired, and he really kind of babied me in a way. My new nephrologist is incredible. **She really started to make me aware of my condition**, the details about it, and what I need to do to take care of myself.*
- age 21

*My mom usually comes with me to appointments and she has all my medical records, but my doctor has always taken care to ask me direct questions. My doctor has also always been encouraging about being able to "live a normal life," and shared that she has Alport patients that have gone abroad for college, and have gotten married. **This gives me confidence about my future**. - age 19*

She didn't patronize me when I was diagnosed at 12, and still doesn't.
I appreciate that. - age 18

Medications

survey results

"I was passing out so frequently that I got to the point when I knew it was coming. I'd sit or lie down and crawl to someplace safe to faint."

"I take Lisinopril and Losartan. I have relatively low blood pressure. When it's hot out, when I sit up or stand up, I feel dizzy and black out."

experience side effects from meds?
79% yes
8% no
13% unsure if symptoms related to meds

side effects experienced:
70% lightheadedness/dizziness
52% tiredness/fatigue
22 % anxiety
22% shortness of breath
17% loss of appetite
4% experienced no side effects

# of medications take daily for Alport syndrome?
57% take 1 medication
17% take 2 medications
27% take 4 or more medications

Experience with ACE/ARB medications interviews

Common Experiences:

Fatigue

Dizziness

Fainting

Challenges waking up

Difficulty with physical activity

I didn't want to take my ACE inhibitor in high school. My nephrologist was after me and pushing me to take it. My parents were on me all the time to take it. When I was 17 years old and on vacation, I googled Alport syndrome and found Alport Syndrome Foundation. ***I started reading people's stories and realized there was a whole community of people living with Alport syndrome****. It was then that I started taking my condition seriously and taking my medication. - age 21*

I take two medications. I have dizziness, fatigue and lightheadedness. *I have to eat a lot and chug a lot of water, and sometimes even just getting up out of my chair I feel dizzy. Working out is always an issue. Change in elevation is a big deal. Almost every other morning after I take my medication, my heart starts racing, I get really thirsty and I can't stop drinking water. - age 17*

I am on 5 mgs of Lisinopril and I also take fish oil. When I was involved in baseball and sports, when I was playing, I didn't experience anything, but as soon as I'd get home, I just wanted to go to bed and I was dizzy. I just couldn't stand up. ***Generally, toward the end of the school day, I find I'm more tired than my friends at school.*** *- age 17*

I tend to feel tired during the day. I have been taking these medications since I was 10 years old, *and the doses have increased. I take them in the morning after breakfast. - age 19*

Psychosocial Aspects

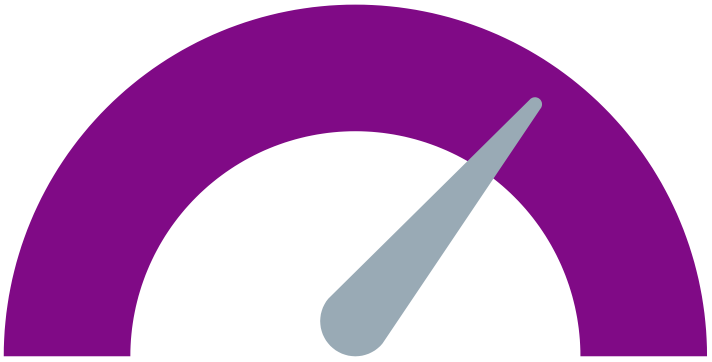
survey results

Have you ever felt alone in your Alport experience?

44% YES

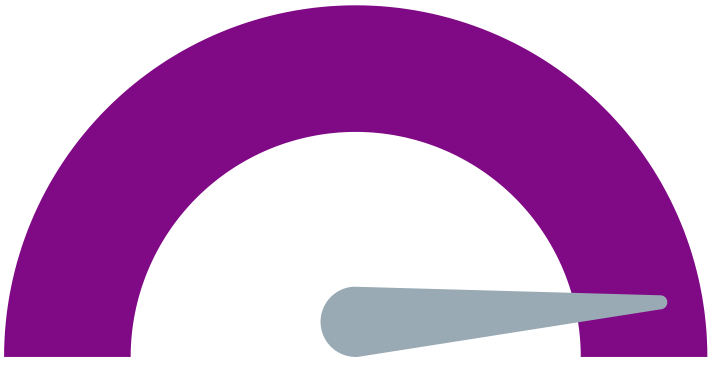
In your daily routine, are you reminded of your disease?

64% YES

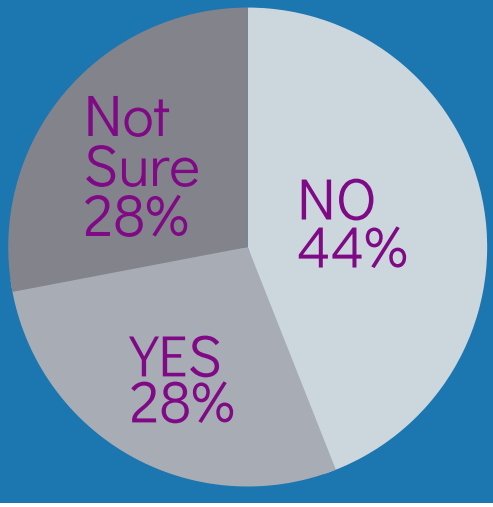


Can you speak with your family about Alport syndrome?

95% YES



Feel your friends understand the challenges you face?



Psychosocial Challenges

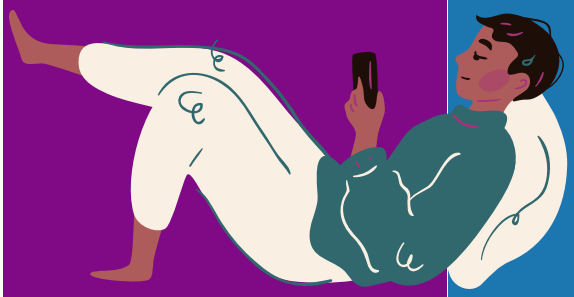
survey results

How much does living with Alport syndrome influence your daily life?

56% moderate to significantly

Most used adjectives to describe daily life with Alport syndrome:

- stressful
- annoying
- time-consuming
- aggravating
- worrisome
- draining
- expensive
- hard



Mental Health & Living with Alport Syndrome interviews

Common Experiences:

Anxiety over potential future renal failure

Concern about having kids someday

Feeling alone

Stress

Post-Transplant Emotions

I still don't know whether I have the recessive or dominant form of Alport syndrome, so there is definitely fear of the unknown about that. When I get in my own head about what's going to be the timeline, what is my prognosis, my roadmap in the future - there is fear, I don't know what that looks like, no one seems to be able to tell me. ***I'm trying to live in the moment and not have to think about it, but when I'm older and starting a family, this is going to become incredibly important.***
- age 20

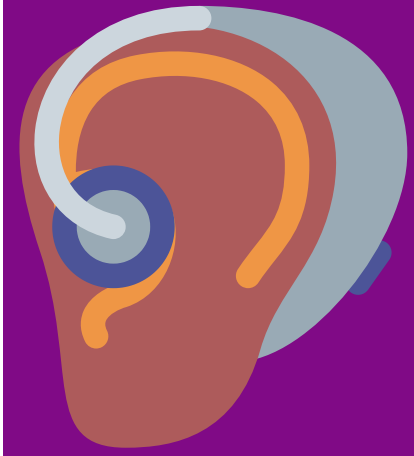
A therapist has been a good constant for me to have in my life. ***If there is one thing that has unlocked the whole Alport syndrome thing for me – it's been talking to other Alport patients.*** *After transplant, I had a lot of emotion. If I wasn't feeling grateful every moment of every day, I felt I was doing a disservice to my living donor. The only thing that helped me through that was talking to other transplant patients and other Alport patients, relating to someone. - age 21*

The stress and anxiety of going to workout. ***I don't want to pass out in front of everyone.*** *Or have some kind of hearing-related thing where I don't have my hearing aids. - age 17*

I was stressed out when I was originally diagnosed. I was concerned about what's going to happen, but it is what it is. ***I've got to move on. I can only control what I can control.***
-age 18

In high school, ***I started doing yoga. That's been helpful to me because it's calming but also aerobic****. It seems good for mental health because it helps calm my anxiety. - age 19*

I have anxiety about renal failure in my future. It has helped to meet people who have gone through transplant and are living happy lives*. I have come to accept who I am and what my challenges are. I used to feel alone but I don't anymore.*
- age 18



Experience with Alport-Related Hearing Loss interviews

Common Experiences:

School anxieties and challenges

Feelings of isolation

Getting used to devices

Hearing deficit fatigue

Ringing in ears

In my junior year of high school, I used an FM system. It wasn't very comfortable and ***the school system didn't really offer the resources to support it well.*** *- age 20*

I was 10 when diagnosed with hearing loss. We had no idea it was related to kidney disease. I was very much a "what, huh?" kind of child and my family was impatient with this for a long time. ***It seemed it was an inconvenience to everyone else that I couldn't hear.*** *No one really considered what I thought or felt about it. It wasn't until my audiologist brought my parents into an appointment with me and shared a computer program that mimics the way I hear things that shifted the way they treated my hearing loss. - age 21*

Fatigue and tiredness are the story of my life*. When you go through kidney failure you definitely experience fatigue. So I don't know if I would be able to identify if I have fatigue from hearing loss. I do have ringing in my ears all the time. - age 21*

My hearing loss is manageable, but there's definitely a lot to manage. Water and sweat with physical activity, especially with the helmet and football. ***Socially, it does affect me in some ways. I guess I'm slowly becoming more isolated.*** *It's not that I can't hear. My hearing aids are great tools. I have moved further and further away from people as they don't relate to it. No one in my family or at my school wears hearing aids. - age 17*

The only time I really think about having Alport syndrome is when people ask about my hearing aids*, or when my hearing aid batteries get low, or when I'm feeling sluggish. - age 18*

I'd rush to get from class to class in middle school to make sure no one saw me drop the FM system off on each teacher's desk. *It was so stressful, and it took up a huge part of my backpack. No one else at my school wore hearing aids. - age 18*

How Living with Alport Syndrome Shapes Me

Common Experiences:

Stressed about future kidney failure

Stress for the family

Striving to live in the moment

Interest in the medical field

Consciously trying not to limit goals

Survey Responses to "How living with Alport syndrome has shaped your experience?"

- 75% stressed about my kidneys failing in the future
- 72% more mindful of my physical health
- 64% more anxious about my future
- 58% causes stress for my family
- 56% more empathetic of others
- 44% brought me closer to my family
- 40% decreased my self-confidence
- 20% have felt hopelessness

I'm studying audiology. *I can connect my own experiences with what I'm learning in class. - age 20*

For two years, I lived day by day watching my labs while they went downhill. Then I had my transplant and everything changed. I'm still figuring out who I am now. ***I think my experience as a patient solidified my interest in becoming a nurse*** *- age 21*

When I step out onto the field, I'm not an Alport patient, I'm a baseball player. *- age 18*

When I was younger, I didn't understand why I was sick and hospitalized. I had to leave school for a semester. The local doctors didn't know what was going on, so they were trying different therapies that made me feel uncomfortable and not confident, in particular when I took prednisone and it made me gain a lot of weight. ***I lost my confidence. Now that I am being treated properly, I am living my life****. - age 19*

I have become an advocate for myself and others with rare diseases. ***I plan to go to college to learn how to become a leader in patient advocacy.*** *- age 18*

After I was diagnosed, I pushed myself to do a study abroad experience. *I thought to myself, I'm just gonna go live in France for four weeks. I love to travel. - age 20*

Perspective to share with newly diagnosed patients

surveys & interviews

- ***Be open about how you're feeling, emotionally and physically,*** *with people you trust and try to take ownership of your disease. I definitely felt empowered once I had the knowledge and confidence to speak with both friends and strangers (through advocacy work) about my experiences.*
- ***Be strong, find some true friends to talk to about it.***
- *Start making* ***healthy habits early.***
- *I would say it's not an easy task living with Alport Syndrome but by keeping a positive mindset you can accomplish anything in life regardless of AS. Don't let it inhibit you from reaching your goals.You can live a normal life even with the complications that come with it.* ***Embrace who you are and don't be afraid to let people know that you have AS, it's a part of who you are!***
- *I would say* ***try to not let it take control of your life.***
- ***Put your medication in a spot or place you won't forget to take it.*** *I put my pills on the kitchen table where I eat every morning so I always remember to take my pills.*
- ***Be grateful for the stage you're at!***
- *Take your medication, it helps. Don't worry about your hearing aids. If people don't like it or jest about it that is on them not you.* ***Just own it. Live your life!***
- *You are different and that's okay.* ***Your normal may be different than other people's normal and that's okay.*** *Embrace that and love yourself because of that, not despite that. Just because you are different than the people around you, doesn't mean you are different from us (ASF members). I didn't have anyone who knew exactly what I was going through and exactly how I felt. I wish I would have found this resource in my early teens.*
- *First and foremost, this is the best possible time to know about your diagnosis.* ***There's a bigger network now than ever before and the science is only getting better. So you're not alone.*** *Secondly, don't be afraid to express yourself. Trying to deal with it yourself only makes it worse.*
- *Take your meds and* ***use it as a motivation not a setback.***

Dr. Bradley Warady

Dr. Bradley Warady is the McLaughlin Family Endowed Chair in Nephrology, Professor of Pediatrics at the University of Missouri-Kansas City School of Medicine. He is also the Director of the Division of Nephrology, and of Dialysis and Transplantation, at Children's Mercy Kansas City.

He currently serves as Co-Principal Investigator of the International Pediatric Peritoneal Dialysis Network and the multicenter, National Institutes of Health (NIH)-funded Chronic Kidney Disease in Children (CKiD) Study. He is also a Council Member for the International Pediatric Nephrology Association (IPNA). He has published more than 300 articles and book chapters. Dr. Warady volunteers at a high level for the National Kidney Foundation, and serves on Alport Syndrome Foundation's Medical Advisory Committee.

ALPORT SYNDROME FOUNDATION®
Hope | Action | Support
www.alportsyndrome.org

Our Resources:

Patients and Families

- Educational Materials
- Network of Support
- Patient and Family Meetings
- Webinars
- Patient Advocacy
- Legislative Advocacy
- Awareness Efforts
- Research Investment

Medical Professionals

- CME/CNE Online (free)
- Diagnosis Information
- Recommended Treatment Guidelines
- Current Research
- Clinical Trial Updates
- Genetics Guide

Researchers

- ASF funds an ancillary Natural History Study in NEPTUNE
- ASF Research Program
- Alport Patient Registry Support
- International Research Collaborations