Diagnosis Stories

ALPORT SYNDROME FOUNDATION®

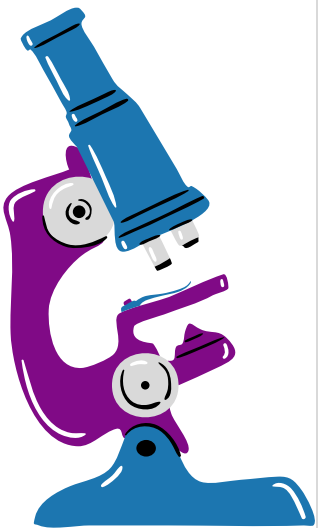
Hope | Action | Support

www.alportsyndrome.org

Survey Results
240 Alport Syndrome
Patients/Caregivers
Self-Reported Responses

February 2021

Contents



Survey Design & Participants

186 Adult Patients
Self-reported responses, age 18+ with confirmed Alport syndrome diagnosis. 69% female. 31% male.

54 First Diagnosed Offspring
Self-reported responses from adults reporting on first offspring with confirmed Alport syndrome diagnosis. 26% female and 74% male offspring.

240 Total Patient Participants

Survey open to all patients age 18+ with confirmed Alport diagnosis and to parents reporting on first offspring diagnosed with Alport syndrome. Invitation to participate promoted through Alport Syndrome Foundation's email and social media communications.

Survey Results Reflect:
January 21 - February 28, 2021.

Participants

Gentic Type: Adults
76% X-linked
10% Autosomal recessive
14% Autosomal dominant

Gentic Type: 1st Offspring
89% X-linked
7% Autosomal recessive
4% Autosomal dominant

Survey questions developed with guidance from our Medical Advisory Committee. Special thanks to Drs: Alessia Fornoni, Rasheed Gbadegesin, Michelle Rheault, Johannes Schlondorff.

Thank you to our project sponsors:

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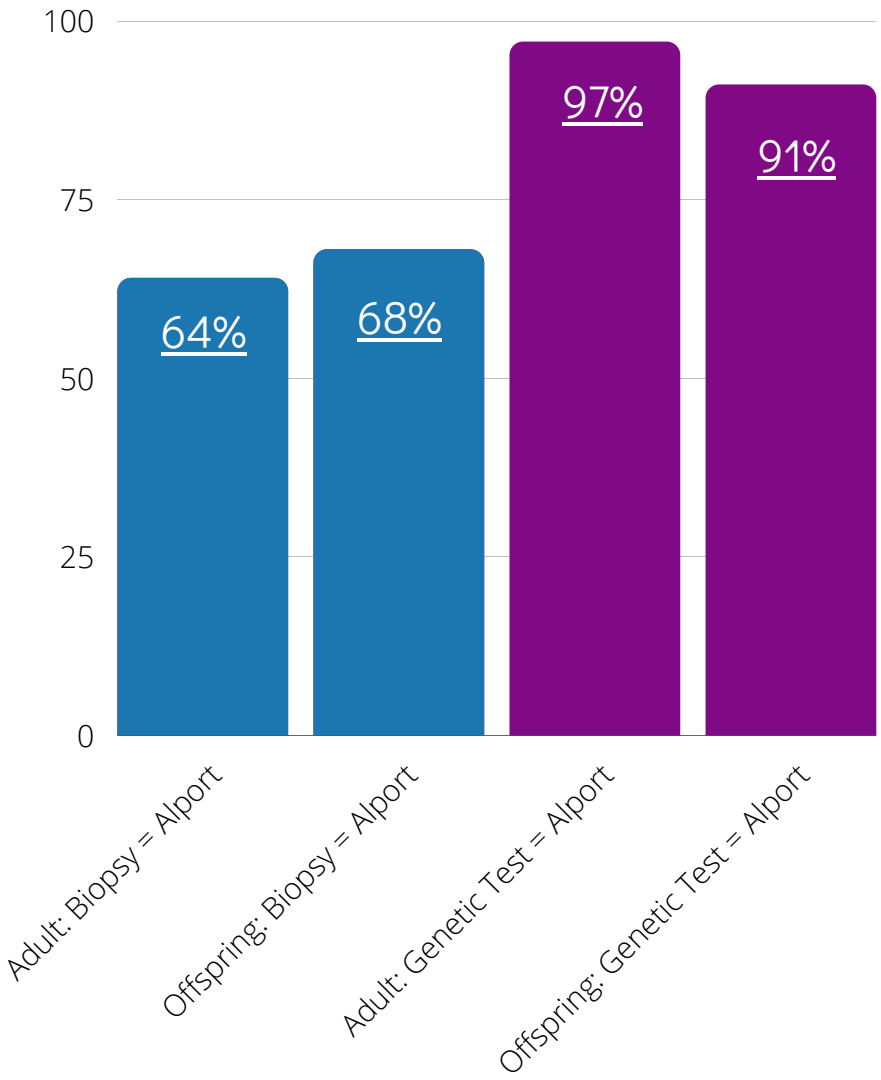
Genetic Testing Impact



57% of adults reported undergoing biopsy, of these patients 36% had results that did not provide accurate Alport diagnosis.
52% of first diagnosed offspring underwent biopsy, of these patients 32% had results that did not provide accurate Alport diagnosis.

68% of adults reported having genetic testing, of these 3% had results that did not provide accurate Alport diagnosis.
83% of first diagnosed offspring had genetic testing, of these, 9% had results that did not provide accurate Alport diagnosis.

Adult Biopsy
111 adults underwent biopsy
71 resulted in Alport diagnosis

1st Diagnosed Offspring Biopsy
28 offspring underwent biopsy
19 resulted in Alport diagnosis

Adult Genetic Test
126 adults had genetic test
122 resulted in Alport diagnosis

1st Diagnosed Offspring Genetic Test
45 offspring had genetic test
41 resulted in Alport diagnosis

Many patients underwent both biopsy and genetic tesing.

Family Impact

"Each of my three children were then diagnosed based on my results."

52% → adult diagnosis led to others in family

This resulted in diagnosis of:

70% siblings

43% parents

41% offspring

A family history of kidney disease helped establish an Alport diagnosis:

51% adults

46% offspring

"My older brother got a transplant at age 5 and had hearing loss. My younger brother got a transplant at 10 and had hearing loss. My mother had a transplant that same year. Six years later I had proteinuria so my doctor ordered genetic testing. Turns out I have Alport syndrome. I'm still at stage 4."

1st person in family diagnosed with Alport syndrome:

53% adults
58% offspring

39% diagnosis of offspring led to others in family

71% led to diagnosis of patient's mother

Misdiagnosis Patterns

51%
of adult patients were initially misdiagnosed or incompletely diagnosed

39%
of 1st offspring diagnosed were initially misdiagnosed or incompletely diagnosed

Received accurate Alport diagnosis 5+ years after renal symptoms were first identified:

47% of adults

18% of offspring

52%
of misdiagnosed adults were told by a physician that they had a benign condition

Mis/Incomplete diagnosis adults:

52% Benign Hematuria/Nephritis

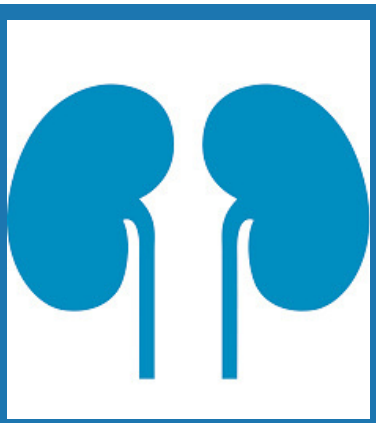
22% Unknown CKD

12% Glomerular Nephritis

12% Thin Basement Membrane Disease

2% Other: FSGS, Cancer, Nephrotic syndrome, UTIs, Minimal Change Disease

"Initial Diagnosis was IgA Nephropathy from 1st biopsy. Changed later to Membranoproferative Glomermulonephritis after 2nd biopsy. After 5 different diagnosis, genetic testing of my daughter resulted in Alport syndrome diagnosis at age 9."



33% of misdiagnosed adults were prescribed medications based on incorrect diagnosis

Symptoms Leading to Alport Diagnosis

primary symptoms

adults/offspring

hematuria: 41% / 50%
proteinuria: 21% / 19%
hearing loss: 11% / 13%

hearing loss

experienced before awareness of kidney disease:

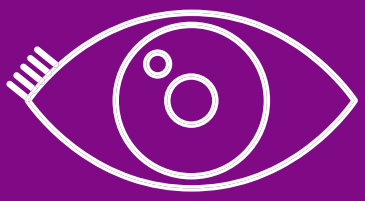
32% adults
19% offspring (1st diagnosed)

blood pressure

experienced before awareness of kidney disease:

34% adults
8% offspring

13% of adults and 6% of offspring experienced eye abnormalities before awareness of kidney disease



11% of adult patients were in renal failure at the time of their diagnosis

Women and Girls with Alport Syndrome

female participants:

120 adults

13 first diagnosed offspring

68% of adult females experienced disease symptoms despite being told by a physician they were "just a carrier" of Alport syndrome

14% of adult females experienced preeclampsia in pregnancy before becoming aware of their Alport diagnosis

54% of female offspring experienced disease symptoms despite being told by a physician they were "just a carrier" of Alport syndrome

"My mother and her sister, myself, brother, two cousins and my niece. Four of us have been transplanted."

"We have been able to trace it back to my great-great grandmother on my mother's side and it has affected seven generations within our family including my two daughters and two grandchildren. I recently celebrated the four-year anniversary of my kidney transplant after spending one and a half years on dialysis."

"My mother was diagnosed at age 16 with chronic kidney disease. Her first transplant at 22, second at 24. My Alport diagnosis at age 40. My daughter diagnosed at 16."

"My father has X-linked Alport syndrome, so we've known for most of my life that I had to be a carrier. Until I attended an ASF family meeting several years ago, I didn't realize that "carriers" could have symptoms too. Since then, I have collected more information to monitor my health."

A Closer Look at the Numbers

Misdiagnosis

Was Alport syndrome your initial diagnosis from a medical professional? Of 186 adult patients that answered this question, 96 answered "no."

Adult patients: If Alport syndrome was not your initial diagnosis, please identify what you were told you had (select all that apply):

28 Benign Familia Nephritis
28 Benign Hematuria
25 Chronic Kidney Disease
13 IgAN or Glomerular Nephritis
13 Thin Basement Membrane
3 Bladder issues/bladder cancer
2 Minimal Change Disease
2 Urinary Tract Infections
1 each: Nephrotic syndrome, Sponge kidneys, high blood pressure, Chronic prostatitis, ruptured spleen, Lupus, Loin pain.

After awareness of kidney symptoms, time it took to gain an Alport diagnosis?
Of 186 adults:
Less than a year: 68
1- 5 years: 30
5-10 years: 13
10 - 20 years: 26
20-30 years: 12
30+ years: 20
Other: 17 (not clear on diagnosis timeline)

Family Impact

97 of 186 adult patients reported their diagnosis led to others in the family. In 40 cases, offspring were then diagnosed. In 68 cases, siblings were then then diagnosed. In 42 cases, parents were then diagnosed. In 15 cases, nieces/nephews then diagnosed. Others also reportedly diagnosed include grandparents, cousins, aunts and uncles.

Symptoms

Prior to becoming aware of kidney issues, did you experience any of the following? Of 186 adult patients that answered this question:

62 - High Blood Pressure
61 - Hearing Loss
25 - Pregnancy Complications
24 - Eye Abnormalities
11 - Esophagus Issues
1 - Aortic Aneurysm

Of 54 first offspring diagnosed:
10 - Hearing Loss
4 - High Blood Pressure
3- Eye Abnormalities
4 - Esophagus Issues

21 - Adults were in renal failure at the time of Alport diagnosis.

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About Us

Our Mission:

To improve the lives of those affected by Alport syndrome through education, empowerment, advocacy and research. Our Vision is to conquer Alport syndrome.

We are a U.S. patient-led non-profit organization educating and giving a voice to the Alport syndrome community.

EIN Non-Profit #20-8237159

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THANK YOU to the patients and families that participated in "Diagnosis Stories" and to the clinicians and researchers who are dedicated to our community.

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