ASF Alport Patient Registry

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Overview

The **ASF** (**A**lport **S**yndrome **F**oundation) **Alport Patient Registry** is an IRB-approved, decentralized, ambispective, longitudinal natural history study launched in August, 2023 and is open to all U.S.-based Alport syndrome patients of all ages with a confirmed diagnosis.

Currently, the registry is available in English. The potential to expand to other languages and/or nations is planned for.

In this poster we highlight some of the unique attributes of this registry which make it an important tool for clinical, translational, and fundamental research in Alport syndrome.

Design

U.S.-based Alport syndrome patients or their pediatric proxies are consented and enter data into a secure web-based platform. Examples of data include:

- Genotype (de-identified patient-uploaded genetic test reports verified by medically & HIPAA trained staff)
- Family history
- pdf/jpg format audiograms showing hearing loss over time
- Disease-specific eye conditions and pregnancy complications
- Standard-of-care medication and treatment tolerance
- Clinical trial participation

Scientific Advisory Board

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Results, Findings, & Additional Perspective

The Registry is built on Pulse Infoframe's healthie™ HIPAA compliant platform, with regulatory-grade, standardized data which allows patients and proxies of pediatric patients to directly enter longitudinal and latest health data through a secure web-based interface. These de-identified data are available for view in aggregate through dynamic visualizations. (*shown right*)

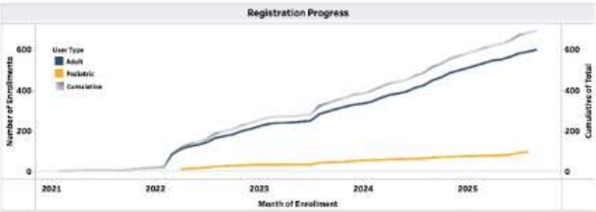
In addition, the patient self-reported data collection approach is superior to an EMR (Electronic Medical Record) for the Alport syndrome patient population specifically because:

- Patient reported data does not contain critical gaps commonly found in EMR records, which would result in lower quality data
- Data tracks with the participant across and independent of clinicians, hospitals, and/or medical systems
- Uploads of pdf format genetic test reports for HIPAA & ACMG compliant source-data verification
- New tool to be added in early 2026 to collect key historical lab values, enriching the data on all participants

Direct feedback through ASF's patient community and medical advisory committee played a vital role in identifying data dictionary elements that position the registry at the forefront of patient-centered drug development. This creates a unique opportunity for collaboration with researchers and sponsors to understand Alport syndrome at a level unavailable elsewhere.

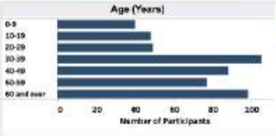
Enrollment

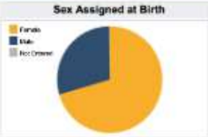
All data and analytics current as of October 11, 2025



ID Survey

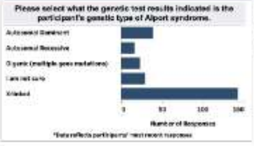
- Age
- Sex at Birth
- Race & Ethnicity *(optional)*

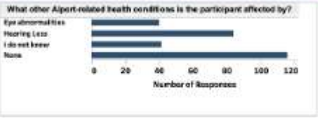


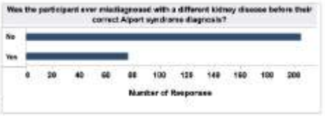


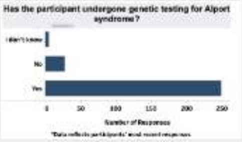
Diagnostic Survey

- Age of Diagnosis
- Means of Diagnosis
- Genotype
- Family History
- Hearing & Eyes
- Audiograms
- ESKD stage
- Dialysis/Transplant
- Lab Values
  - Creatinine
  - eGFR
  - Cystatin C
  - uACR
  - K+
- Height & Weight *(<18yo)*
- Pregnancy









Life & Medication Survey

- Diet
- Smoking *(>18yo)*
- Drinking *(>18yo)*
- RASi
  - ACEi
  - ARB
- SGLT2i