

ASF Alport Patient Registry



Authors

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Introduction

The ASF (Alport Syndrome Foundation) Alport Patient Registry is an IRB-approved, decentralized, ambispective, longitudinal natural history study launched in August, 2023 and is open to all Alport syndrome patients with a confirmed diagnosis.

Currently, this registry is available to Alport syndrome patients in the United States.

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

Design

- Patients in the U.S. who have Alport syndrome are consented and enter data into the platform. Examples of data include:
- Clinical validation of de-identified patient uploaded genetic test reports to verify genetic type(s) and variant(s) by medically trained staff
 - Pdf/jpg format audiograms showing hearing loss over time
 - Disease-specific eye conditions and pregnancy complications
 - Standard of care medication and treatment tolerance

Direct feedback through ASF’s in-person and virtual patient meetings, surveys, and private online support group played a vital role in identifying data elements that position the registry at the forefront of patient-centered drug development in Alport syndrome.

Analysis

The latest* enrollment and statistics related to diversity of ages, stages of disease, ethnicity, genetic types and more are shown below.

* Current as of October 7, 2024.

Data dictionary question examples

ID Survey

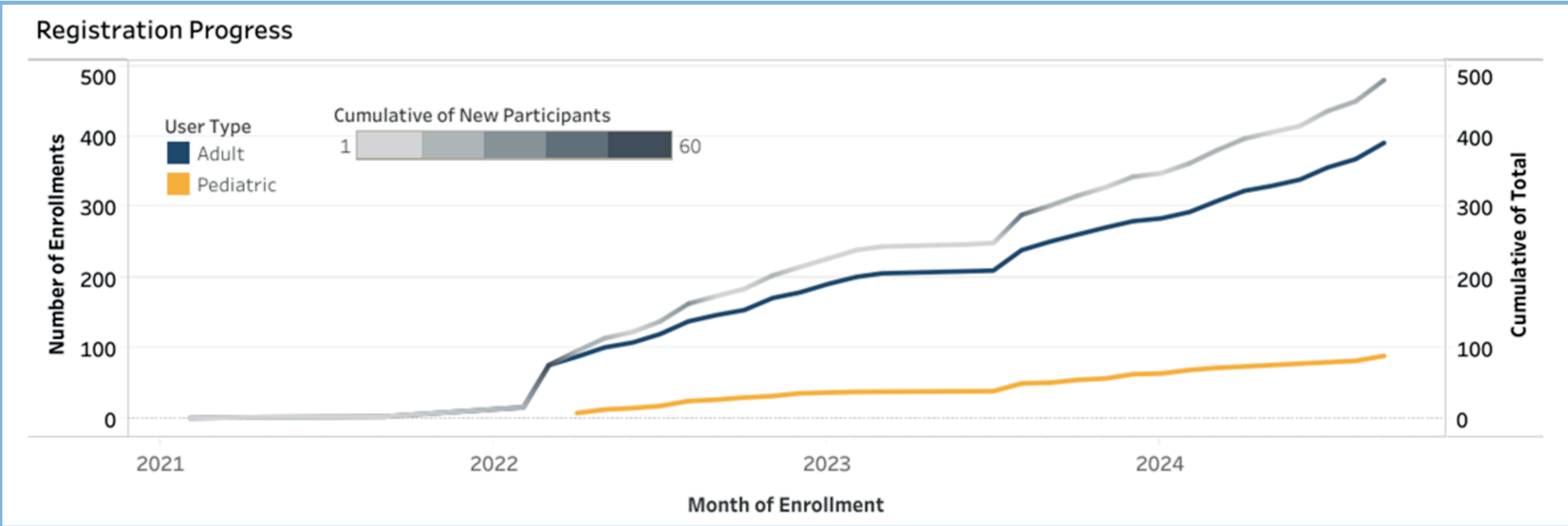
- Sex at birth
- Age
- Race & Ethnicity (optional)

Life & Medication Survey

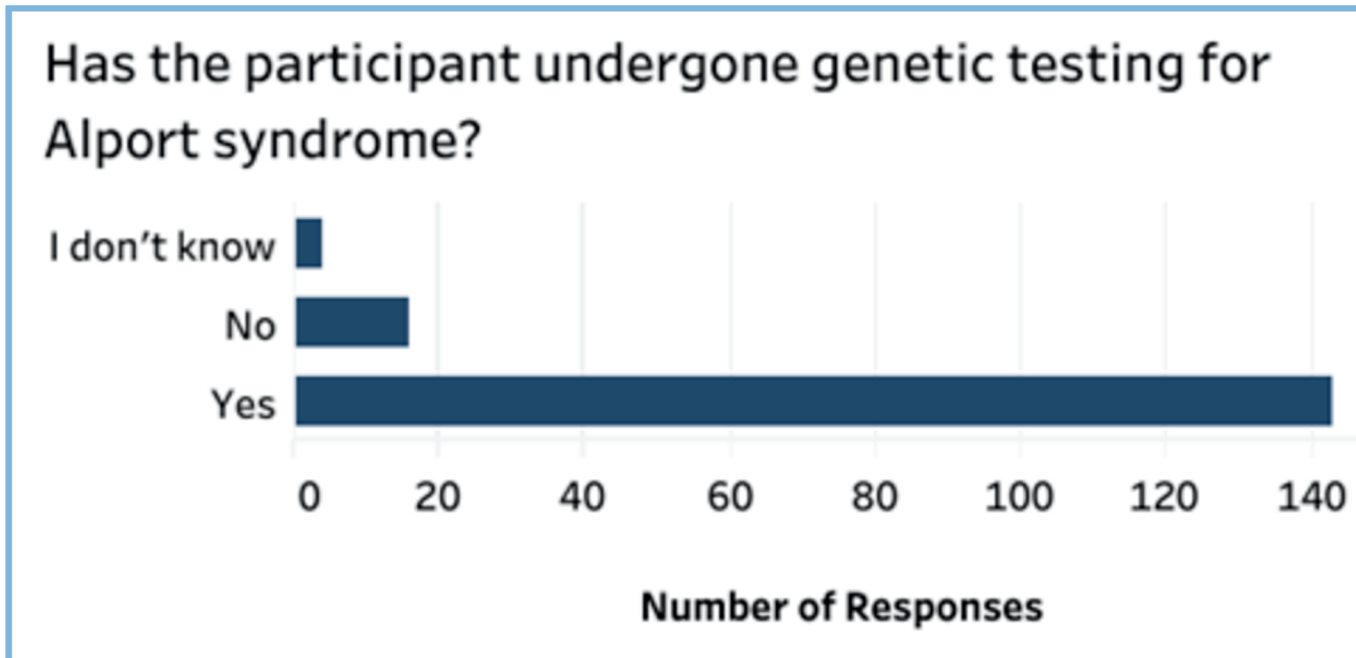
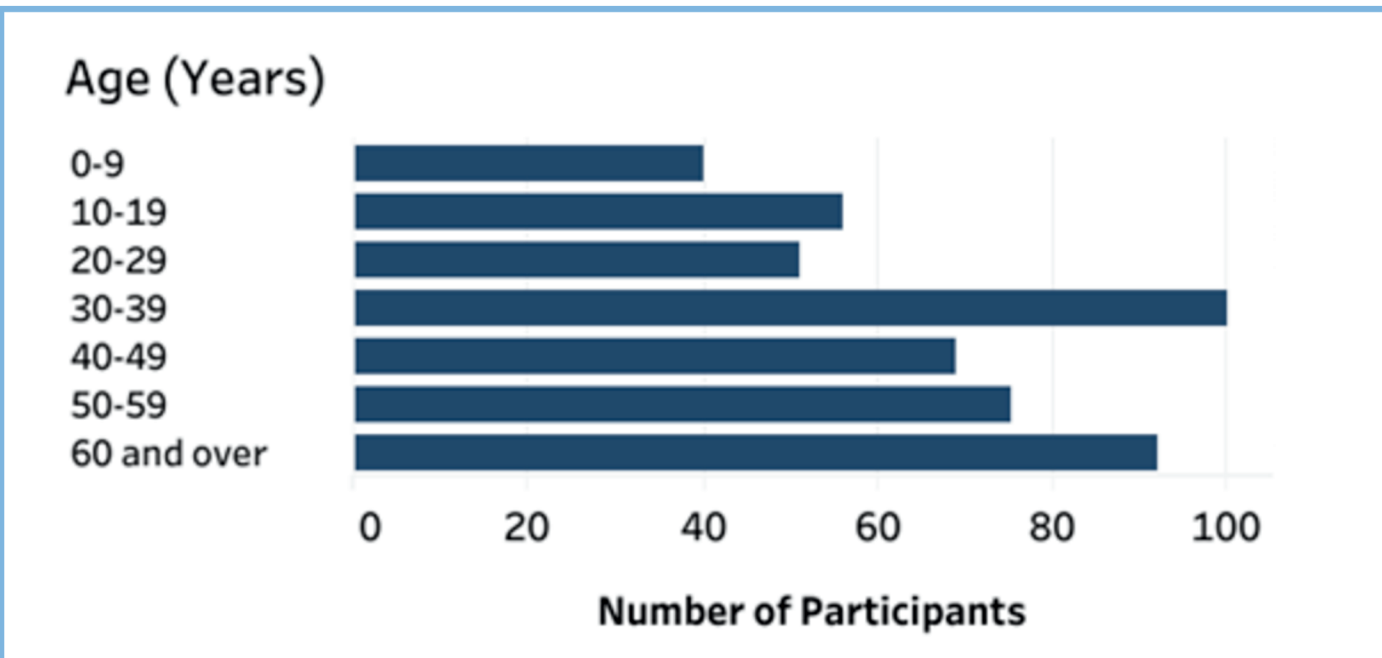
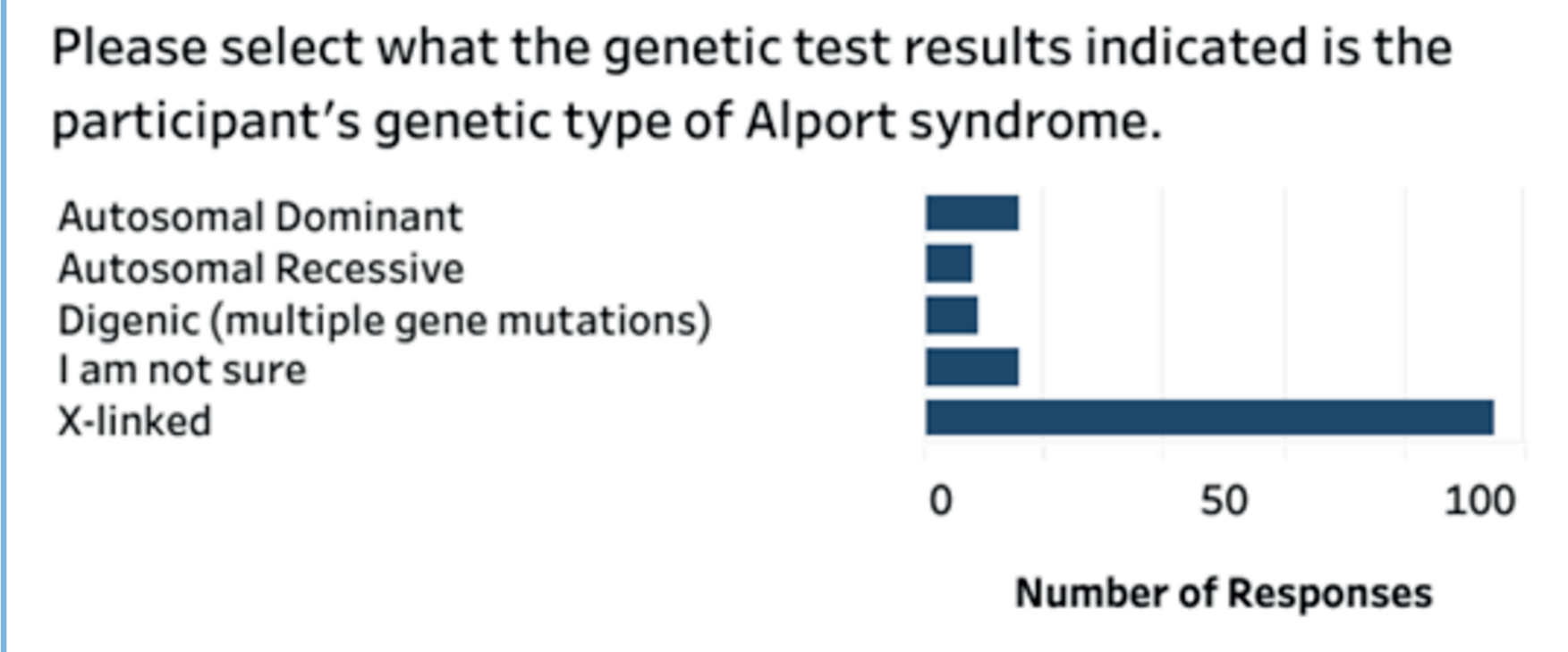
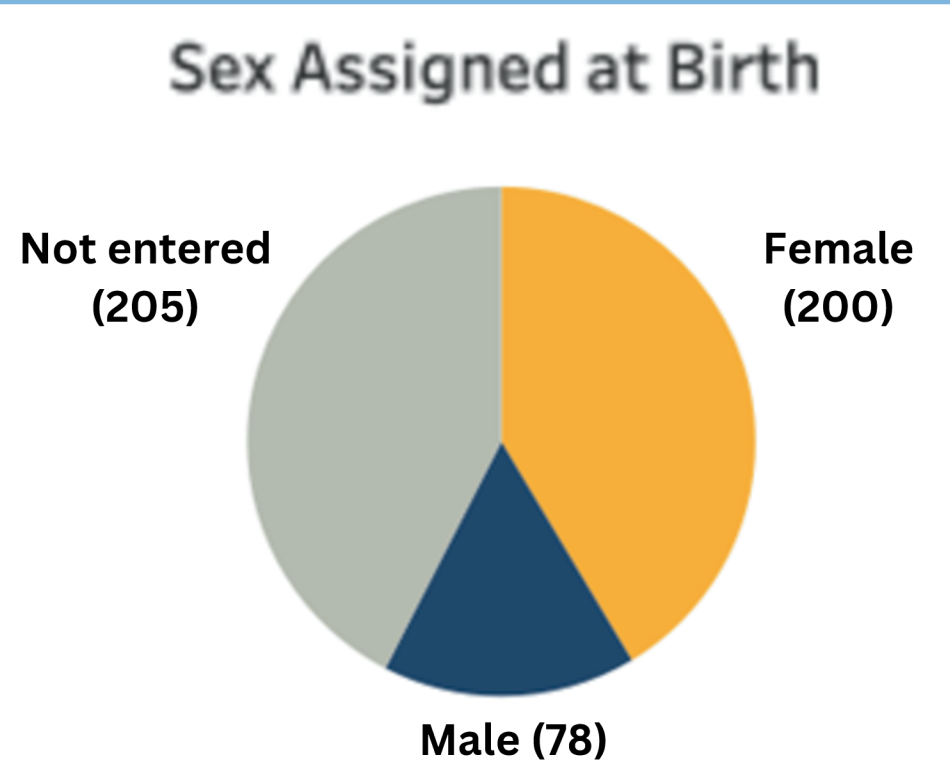
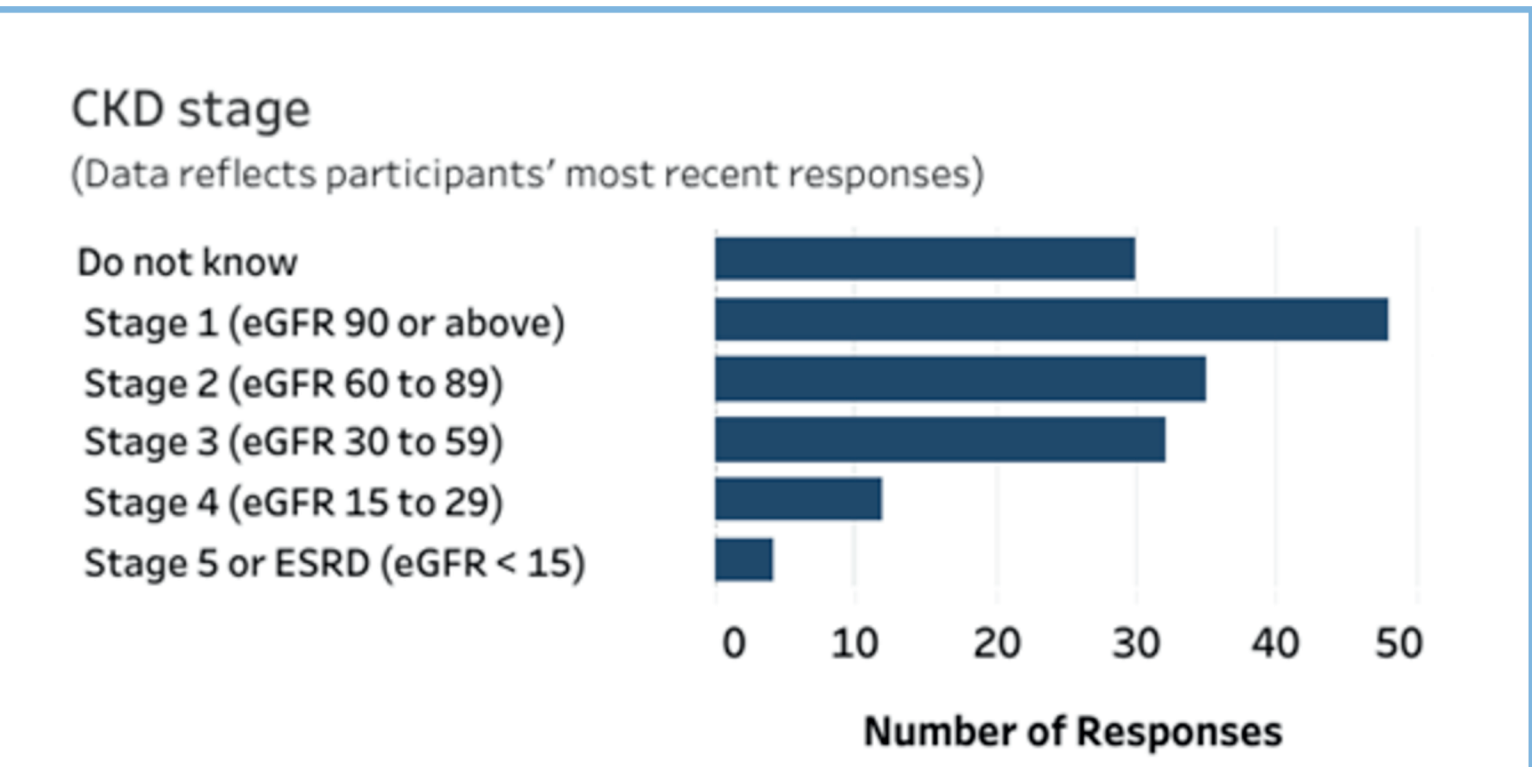
- Smoking (>18yo)
- Drinking (>18yo)
- RASi Treatment History
- SGLT2i Treatment History

Diagnostic Survey

- Age of Diagnosis
- Means of Diagnosis
- Genetic test report
- Family History
- Hearing & Eyes
- Audiograms
- ESKD stage
- Dialysis/Transplant
- Lab values: (eGFR, Creatinine, K+, UACR)



Race		Ethnicity	
Asian	4	Hispanic or Latino	23
Asian Indian	3	Not Hispanic or Latino	257
Black or African American	3	Prefer not to answer	4
Native Hawaiian or Other Pacific Islander	1	Unknown	4
Prefer not to answer	2	Not Entered	195
White	129		
Unknown	1		
Not Entered	340		



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Results/Findings

The Registry is built on Pulse Inframe’s healthie.net HIPAA compliant platform, with regulatory-grade, standardized data which allows patients and proxies of pediatric patients to directly enter health data through a web-based interface.

These data are available for view in aggregate through dynamic visualizations.

The platform offers interactivity and ease of use for participants, ongoing record collection every six months to monitor disease progression/symptoms/medication changes, and a library of educational resources for patients. This creates a unique opportunity for collaboration with researchers and sponsors.

Additional Perspective

- There is future flexibility to offer participation in different languages. In addition, the patient self-reported data collection approach is superior to an EMR (Electronic Medical Record) for the Alport syndrome population specifically because:
- Patient reported data does not contain critical gaps commonly found in EMR records, which would result in lower quality data
 - Patients have less trust in EMR data as a result, and have expressed a preference for a patient-reported approach

With ASF, a patient-led and patient-focused organization, as the sponsor of the registry, its collaboration with Pulse Inframe should provide an ideal combination to generate quality, real-world medical data in a secure and research-friendly format.