



## **Alport Syndrome Patient Volunteers Needed!**

### Human Gene Modifier Data Study

This is part of a larger research project funded in 2022 by The National Institute for Diabetes and Digestive and Kidney Diseases. Dr. Ron Korstanje at The Jackson Laboratory in Bangor, Maine is the Principal Investigator of the study. The research compares gene modifiers in mouse models with those found in human patients affected by Alport syndrome. The goal is to help support better understanding of potential effective pathways for drug therapy in Alport syndrome.

The Co-Investigator for the patient volunteer aspect of the study is Dr. Laith Al-Rabadi, who serves on the [Medical Advisory Committee](#) for Alport Syndrome Foundation. Dr. Al-Rabadi is both a clinician that treats patients, and a researcher at the University of Utah's School of Medicine. Dr. Al-Rabadi welcomes patient questions as well and can be reached at [Laith.Al-rabadi@hsc.utah.edu](mailto:Laith.Al-rabadi@hsc.utah.edu).

**For more information and/or to volunteer as a participant, contact the study coordinator: Autumn Wilcox at University of Utah: [Autumn.Wilcox@hsc.utah.edu](mailto:Autumn.Wilcox@hsc.utah.edu)**

### Volunteer participation in the study includes the following examples:

- Sharing some of your basic renal laboratory results (typical metabolic panel blood tests) with the study coordinator via mail or email.
- Accepting and using a saliva kit sent by mail to your address and shipping back to the study site in a pre-paid package.

### Volunteer requirements:

- Confirmed diagnosis of Alport syndrome (through biopsy, skin and/or genetic test).
- Age 7 or older.
- Patients living in any country are eligible.
- Patients with all genetic types of Alport syndrome are eligible (also patients diagnosed with thin basement membrane disease).

All patient data will be deidentified in research documents, reports, and potential publications. The goal of the study is to collect data from a minimum of 100 patients.