Dr. Ron Korstanje, a member of Alport Syndrome Foundation’s Scientific Advisory Research Network, leads a research team at The Jackson Laboratory (JAX) that was recently awarded a four-year grant from the National Institute of Diabetes and Digestive and Kidney Disease totaling $1.49M to study Alport syndrome genetics and identify modifier genes affecting the severity of disease and age of onset. This major grant allows Dr. Korstanje to expand the work he began with an initial award of $125,000 from Alport Syndrome Foundation (ASF).

“Our studies will lead to a better understanding of the initiation and progression of the disease process in general,” Korstanje says, “and will help to reveal novel therapeutic targets to combat end-stage renal disease in patients with Alport syndrome as well as patients with other forms of kidney disease.”

The project, which will study both mouse and human models, is a collaboration with Dr. Laith Al-Rabadi (University of Utah). Dr. Al-Rabadi, a clinician and researcher who serves on Alport Syndrome Foundation’s Medical Advisory Committee, will test in human patients for the candidate genes that Dr. Korstanje and his team identify in the mouse models.

“The current proposal will use whole exome sequencing and targeted genetic testing in patients with Alport syndrome and their family members. We will carry out an approach to identify inter and intra-family variation in a focused and streamlined manner for a classically inherited syndrome. We will use the data we generated from mice for suspected modifier genes and validate it in a large patient cohort. This field will only advance by active patient participation,” notes Dr. Al-Rabadi. Patients interested in assisting Dr. Al-Rabadi can contact study coordinator Autumn Wilcox at Autumn.Wilcox@hsc.utah.edu. A blood or saliva sample and clinical information are all that is required to participate.

Alport syndrome is an inherited disease characterized by progressive decline in kidney function, hearing loss, and eye abnormalities. The disease is caused by genetic mutations that affect the expression or function of the type IV collagen α345 family of proteins in the basement membranes of affected organs. Estimated to affect less than 200,000 people in the United States, Alport syndrome is considered to be a rare disease. Currently, there is no FDA-approved therapy for Alport syndrome.

Led by and for patients, Alport Syndrome Foundation’s mission is to improve the lives of Alport syndrome patients through education, empowerment, advocacy, and investment in research. ASF directs the majority of its resources to research-related activities and patient education. Since it was established in 2010, ASF’s Research Program has invested more than $2M in research projects globally. To date, that investment has led to more than $20M of additional investment from other sources to expand the initial efforts of ASF research award recipients.

Lindsey Avery Fitzsimmons, an incoming postdoctoral fellow who will be working closely with Dr. Korstanje at The Jackson Laboratory on this project, is a kidney patient herself. “This is a pivotal moment for me, not only in my professional career and scientific training, but also as a patient only recently diagnosed with a rare, genetic form of chronic kidney disease,” she said. “It’s an incredible feeling to be awarded such an opportunity that I hope will yield informative and meaningful results to maximally improve our understanding of Alport syndrome processes and streamline the development and implementation of future therapeutics.”

Alport syndrome patient and ASF Executive Director Lisa Bonebrake notes, “ Funds for our Research Program are provided by patients, families, and friends. It is impactful that seed funding from ASF assists dedicated Alport syndrome researchers in securing additional funding to expand and sustain their efforts. We are grateful to Drs. Korstanje and Al-Rabadi for their continued work in Alport syndrome.”

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