



## GIGA-Kids: Tracing the Genetic Roots of Childhood Kidney Disease

### What Is IgA Nephropathy?

- IgA nephropathy is a form of kidney disease that occurs when IgA, a protein that normally helps the body fight infections, collects in the small blood vessels of the kidney. This causes inflammation and scarring of the glomeruli, the kidney's tiny filtering units.
- Over time, the result is kidney malfunction. More than 20 percent of IGAN patients eventually experience kidney failure and require a transplant.
- Previous research has shown that genetic makeup plays a key role in who develops IgA nephropathy. Further research in genomics will tell us much more about how IGAN works and may advance our understanding of other forms of kidney disease as well.

**GIGA-Kids** is a collaborative research effort to understand the genetic basis of IgA nephropathy (IGAN), a common form of kidney disease that is the leading cause of kidney failure among young adults. With support from the IGA Nephropathy Foundation of America and the Midwest Pediatric Nephrology Consortium, Columbia University Medical Center researchers are leading a genomic study of IGAN in pediatric patients. Known as GIGA-Kids (or the Genomics of IgA-related diseases in Kids), this study will shed valuable light on the genetic and biological factors underlying IgA nephropathy.

### Recent Progress in IGAN Genomics

Researchers at Columbia University Medical Center recently completed a major genomic study of more than 20,000 adults with IgA nephropathy. The study identified specific regions of the human genome associated with IGAN, and in the process, uncovered a surprising link between IGAN and the intestine. IgA nephropathy, it turns out, may actually be an inflammatory disease that begins in the intestine. This insight could lead to new therapies based on current treatments for inflammatory bowel disease, and suggests new biological pathways to explore for pharmaceutical development.

### Implications for Children With IGAN

Since IgA nephropathy is often not diagnosed before patients are very sick, the ability to intervene earlier—and to understand how the disease progresses in young patients—is of paramount importance. Columbia's recent research has identified a number of genetic risk factors for IGAN in adults, as well as a key biological mechanism at play in the early stages of disease. It is unclear to what extent these discoveries apply to pediatric patients, but the ability to improve diagnosis of IGAN in children would be groundbreaking as it could allow treatment to begin before the kidneys are damaged. GIGA-Kids will pursue this goal by testing for the newly-discovered biomarker and genetic risk factors in children. The results will provide a great advance to our understanding of how IGAN leads to kidney failure and what we may be able to do to stop it.

## World-class Expertise

### *Krzysztof Kiryluk, M.D.*

Dr. Kiryluk is the Marc Anthony Zambetti Assistant Professor of Medicine at Columbia University Medical Center. A specialist in chronic kidney disease, glomerulonephritis, and inherited kidney disorders, Dr. Kiryluk leads several large collaborative national and international genetic studies of glomerular disorders.

### *Ali Gharavi, M.D.*

Dr. Gharavi is chief of the Division of Nephrology at Columbia University Medical Center. His research is focused on the molecular genetics of kidney diseases, particularly glomerular and developmental disorders. The long-term goal of his research is to identify the specific genes and biological pathways underlying kidney disorders in order to facilitate the development of new diagnostic tools and therapies.

### *Midwest Pediatric Nephrology Consortium*

The Midwest Pediatric Nephrology Consortium is a group of leading medical research institutions working to improve care for pediatric nephrology patients. The consortium is taking a lead role in enrolling patients in GIGA-Kids, and in the collection of biological specimens, clinical data, and patient and family histories for the study. These efforts are led by two international experts in pediatric kidney disease: Dr. Robert Wyatt from Le Bonheur Children's Hospital in Memphis and Dr. Raoul Nelson from the University of Utah.

## Championing Kidney Health



### *Donald Jones, Former NFL Wide Receiver*

Donald Jones was diagnosed with IgA nephropathy during his sophomore year of college. He went on to play wide receiver for three years in the National Football League with the Buffalo Bills, and briefly with the New England Patriots, before he experienced renal failure and was forced to retire early. Jones was treated at Columbia University Medical Center, where he received a kidney transplant from his father.

After a successful recovery, Jones has committed himself to community education, working to raise awareness of health issues, in particular kidney disease. He is currently extending his support to the GIGA-Kids study.

## The Future and How You Can Help

The GIGA-Kids study has been made possible by generous support from the IGA Nephropathy Foundation of America, but further financial support is still needed to complete this important research.

If you would like to get involved or learn more about IgA nephropathy, please contact Bonnie Schneider at the IGA Nephropathy Foundation of America at (732) 770-7377. You may also visit [www.iganephropathy.org](http://www.iganephropathy.org).

For more information about supporting the GIGA-Kids study or to make a gift, please contact Aislinn White, Director of Development at Columbia University Medical Center, at [aislinn.white@columbia.edu](mailto:aislinn.white@columbia.edu) or (212) 342.3147. You may also visit [www.gigakids.org](http://www.gigakids.org).