

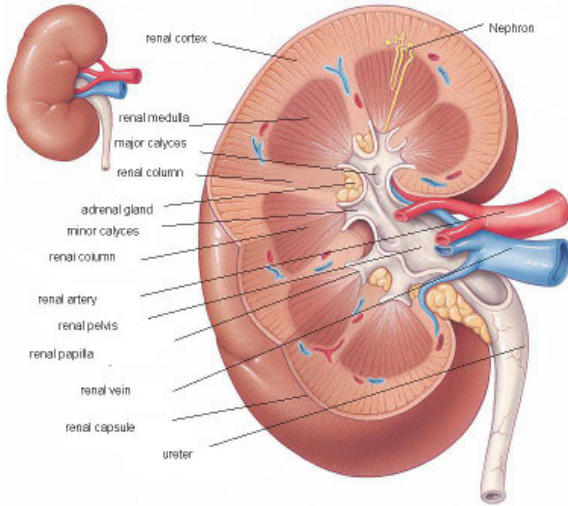
Genetics of Kidney Disease Research Update

Fall 2017

Vol. 1 Issue 1

Welcome to News about the Genetics of Kidney Disease Research!

This is the first newsletter for families with Kidney Disease produced by the Duke Molecular Physiology Institute (DMPI) at Duke University Medical Center (DUMC). The genetics of kidney disease research team at DMPI includes a team of dedicated research professionals and scientists looking for the unknown genetic causes of different kidney diseases. Our team is led by Dr. Rasheed Gbadegesin, a Pediatric Kidney Doctor at Duke University Hospital.



What is Kidney Disease?

The kidneys are important organs that help filter waste products from our body. When the kidney is not working properly, there is accumulation of waste products and excess water in the body. We are currently studying several specific diseases including:

- Nephrotic Syndrome (NS) – symptoms include protein in the urine, body swelling, and low amount of albumin in the blood.
- Focal Segmental Glomerulosclerosis (FSGS) – Is a type of nephrotic syndrome resulting from scarring of the kidney filters. It is an important cause of kidney failure in adult and children especially in black people.
- Vesicoureteral Reflux (VUR) – urine flows “north” back toward the ureters and kidney instead of “south” out of the body. Affected individuals are more prone to urine infection. If VUR is not detected early, it can cause scarring of the kidney.

Enrollment Update

Since opening recruitment, we have enrolled over 2100 patients and family members into this study from around the world! We could not do this research without the great partnerships that we have with our patients and their families. Thank you for your interest and/or participation!

Recruitment is on-going and we hope to eventually enroll 3000 people into our study. If you know anyone who might be interested in participating, please ask them to contact the study team—they do NOT need to be Duke patients to be eligible.

Recruitment Contact:

Lynn Rodgers
Phone: 919-681-2079
Lynn.rodgers@duke.edu

Why are we researching Kidney Disease?

As Kidney Disease advances, it can lead to Kidney Failure which is often not reversible. Kidneys that no longer work right, can cause increased swelling or fluid build-up in the body and harmful wastes increase in the blood. Treatment can be limited to dialysis, where the body is hooked up to a machine that works as an artificial kidney, or transplant, where the patient needs a donor kidney from someone else. Unfortunately, thousands of people will die in the U.S. every year from kidney failure.



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Important terms to know:

- **DNA**—deoxyribose nucleic acid which is strands of genes twisted together.
 - **Gene**—a tiny structure in every cell that is the blueprint for our body. Genes come in pairs and make up chromosomes.
 - **Mutation**—a permanent change in a gene that is different from normal.
 - **Nephrotic Syndrome (NS)**—a condition of the kidneys which can lead to kidney failure.
 - **Focal Segmental Glomerulosclerosis (FSGS)**—a type of NS where scarring inside the kidney that affects how the kidney works.
- Edema**—extra fluid that builds up in the body when the kidneys don't filter the blood normally.
- **Proteinuria**—abnormal protein in the urine.
 - **Albumin**—the most common type of protein in the blood.
 - **Excrete**—to separate or expel waste from the body and the main function of the kidneys.
 - **Hypertension**—a condition when the fluid in the body's arteries increases; also known as "high blood pressure."
 - **Vesicoureteral Reflux (VUR)**—a condition where urine flows backwards from the bladder up through the ureters and into the kidney.
 - **Ureter**—a tube that connects the bladder to the kidney
 - **Urinary tract**—includes the kidneys, ureters, bladder, and urethra.
 - **Voiding Cystourethrogram (VCUG)**—a medical procedure that uses a special x-ray technique to look at the function, size, and shape of the urinary tract.

Meet the Research Team!



Rasheed Gbadegesin, MD, is a Pediatric Nephrologist and leads our study. He works tirelessly to not only see his patients in clinic and in the hospital, but to run the research lab. He has been with Duke for 8 years after training in Nigeria, United Kingdom and the USA.

Megan Chryst-Ladd is a Research Technician in Dr Gbadegesin's laboratory.



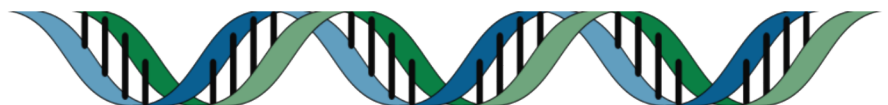
Lynn Rodgers is the research coordinator for our study. If you have any study-related questions, she is happy to help.

Other key personnel of our study include:

The Pediatric Nephrology team of , Dr. Delbert Wigfall, Dr. Shashi Nagaraj, Dr. Annabelle Chua, Dr. Eileen Chambers and Nurse Practitioner Lisa Patterson.

The Pediatric Urology team of Dr. John Weiner, Dr. Jonathan Routh, Dr. Todd Purves and Nurse Practitioner s Cindy Camille and MaryEllen Kelly.

And last, but most definitely not least, our laboratory technician extraordinaire, Megan Chryst-Ladd. We absolutely could not have had the success that we have had without her.

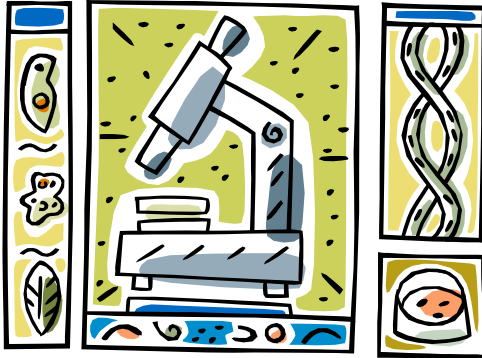


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Groundbreaking News!



In April 2013, the *Journal of the American Society of Nephrology* (JASN) published our paper about our latest findings on hereditary Vesicoureteral Reflux. We identified a defect in a gene called *TNXB* in a large family with both joint hypermobility and vesicoureteral reflux. We suspect that the defect or mutation that we found in *TNXB* can also cause mechanical problems at the location where the ureter meets the bladder. We are still exploring the connection between joint hypermobility and VUR.

In March 2014, we reported a new mutation in a gene called anillin (*ANLN*) as a new cause of Focal Segmental Glomerulosclerosis (FSGS) in a family with many affected individuals. Defects in *ANLN* resulted in FSGS by abnormal movement of a cell in the kidney called the "Podocyte." We published this paper in JASN as well. We are currently exploring other ways by which this defect will cause FSGS and kidney failure.

In 2015, we identified common genetic factors that may predispose children to a type of nephrotic syndrome called Steroid-Sensitive Nephrotic Syndrome (SSNS). We identified defects in the gene of the immune system, which if present in combination with other factors in the environment, may predispose patients to nephrotic syndrome. There are probably hundreds of these factors and we are still searching for these additional factors in other children. We published our findings on this in the July issue of JASN. SSNS is the most common variety of NS, affecting greater than 80% of those children and results in patients who can respond to steroid (standard) therapy. Further testing is on-going.



Vesicoureteral Reflux Research Update

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Word Search

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Doctor	Potassium
Duke	Proteinuria
Edema	Reflux
Electrolytes	Renal
ESRD	Research
Excrete	Science
Gene	Transplant
Glomeruli	Ureter
Healthy	



Our most sincere thanks to you for your interest and participation in this study. Research is only possible with the cooperation of our patients and we hope that you are as excited as we are about the future implications of our findings. Please stay tuned for the next update!

Dr. Gbadegesin and team

