5 Rare Kidney Diseases *You May Not Know About*

FAMILY FEATURES

While the leading cause of kidney disease is diabetes, many other factors can lead to kidney disease and failure – including a collection of rare and genetic conditions. According to the National Organization for Rare Diseases (NORD), a disease is considered rare if it affects fewer than 200,000 people in the United States. Today, 30 million Americans are living with rare diseases.

This Rare Disease Day, observed on Feb. 28 worldwide, the American Kidney Fund is committed to improving the understanding of rare kidney diseases by providing educational resources.

IgA Nephropathy

An autoimmune disease, IgA nephropathy (IgAN) is related to improper function of the immune system. IgAN causes the immune system to produce abnormal antibodies, which build up in the kidneys, triggering inflammation and reducing the kidneys' ability to filter waste and fluid, causing damage and potentially leading to kidney failure.

According to NORD, approximately 70% of rare diseases begin in childhood, which was the case for Malkia White. She had no symptoms – the only indication of her kidney problem was protein and blood in her urine detected through a routine test. She was diagnosed with IgAN but continued living her life without any changes – the disease was so rare, little was known at the time about how to manage it.

"From 6 years old to the age of 42, I maintained my medical appointments and lived an active lifestyle," White said. "I was an honor student. I was always in dance class. In high school, I was in a marching band and on the field hockey team. In that time period, I was being checked. It never occurred to me, or my family, to investigate or research [IgAN]."

APOL1-Mediated Kidney Disease

Known as AMKD, this is a spectrum of kidney diseases associated with variants (mutations) in the apolipoprotein L1 (APOL1) gene. Everyone has two copies of the APOL1 gene, but mutations of the gene can raise the chance of rapidly progressive kidney disease in people of western and central African descent.

Polycystic Kidney Disease

Polycystic kidney disease (PKD) is a genetic disease that causes cysts to grow inside the kidneys. There are two forms of PKD: autosomal dominant polycystic kidney disease (ADPKD) and autosomal recessive polycystic kidney disease (ARPKD). The former is more prevalent, accounting for about 9 of 10 cases of PKD.

Cystinosis

A rare, multisystem genetic disease, cystinosis accounts for nearly 5% of all childhood cases of kidney failure, although some people with cystinosis do not develop kidney disease until they're teens or adults. Caused by mutations in the CTNS gene, cystinosis happens when cystine, a component of protein, builds up in your body's cells. Too much cystine causes crystals to form and can damage organs including kidneys, eyes, pancreas, liver and brain.

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Complement 3 Glomerulopathy

With complement 3 glomerulopathy (C3G), a part of the immune system called the complement system becomes overactive and doesn't work properly, leading to damage and inflammation in the kidneys. Specifically, it damages the kidneys' glomeruli, which help kidneys filter toxins out of the blood. It can cause kidney failure in about half of adults who are diagnosed with the disease.

Michelle Farley had a hard time getting her C3G diagnosis despite high blood pressure and an irregular heartbeat in her youth and suffering from daily vomiting and weekly headaches while in college. After a trip to her college medical center, she discovered her blood pressure was so high she was at risk for stroke or heart attack. Bloodwork determined she had markers for kidney disease, but she wouldn't receive a full diagnosis until she was 25.

"I was left undiagnosed for almost 22 years due to preconceived notions of how disabilities and sicknesses should 'look' on the outside and how old you need to be to have a chronic disease," Farley said. "I think it's important to spread awareness about rare kidney diseases so patients can be diagnosed faster and more accurately. I always wonder how long I could have maintained my native kidneys if I was diagnosed as a child."

Learn more about rare kidney diseases and the Rare Kidney Disease Action Network by visiting kidneyfund.org.