FAST*facts*

June 2026

WHY IDENTIFYING THE CAUSE OF RARE KIDNEY DISEASE MATTERS

Rare kidney diseases carry an outsized burden. Each condition may affect only a small share of the population, but together they cause a striking number of kidney failures, often in children and young adults.

For a young person whose kidney function is declining, or anyone whose family has a history of kidney disease, identifying the cause is what unlocks the right treatment and changes the course of the disease. Below are fast facts on rare kidney disease and why getting to the root cause matters.

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Q: What are rare kidney diseases?

Rare kidney diseases are a subset of chronic kidney disease, a global health crisis that affects more than 10% of the world's population and causes more than 2.6 million deaths each year.[1,2]

A disease is considered "rare" when it affects fewer than one in 2,000 people.[3] More than 500 kidney diseases meet that threshold.[4] Some conditions in this group, such as polycystic kidney disease and IgA nephropathy, are more prevalent than most rare kidney diseases but are still included because they share key features with rarer conditions and are not caused by common cardiometabolic conditions like diabetes and hypertension. Other conditions like C3 glomerulopathy (C3G) and atypical hemolytic uremic syndrome (aHUS) are considered ultra rare.

Together, rare kidney conditions cause a large share of kidney failures worldwide. Kidney failure cuts life expectancy sharply and costs patients years of productive life, even with dialysis or a transplant.

> Rare kidney diseases account for a large share of kidney failure.

Q: What causes rare kidney diseases?

Rare kidney diseases have many causes, and many of them are still poorly understood. Genes play a big role. Mutations in a single gene cause up to half of childhood kidney disease and about 10% of adult cases.[5] Variants in a gene called APOL1, more common in people of African ancestry, also raise the risk. Examples of genetic, rare kidney diseases include Alport syndrome and polycystic kidney disease.

> Most causes of rare kidney disease are poorly understood.

The immune system is another major driver. In autoimmune kidney diseases, the body's defenses turn on its own tissue — sometimes set off by genes, environmental exposures or conditions like lupus. In many cases the cause is never identified, and some of those "unknown" cases are likely undiagnosed rare kidney diseases. Common immune-related examples include IgA nephropathy and lupus nephritis.

Q: How do rare kidney diseases harm young people?

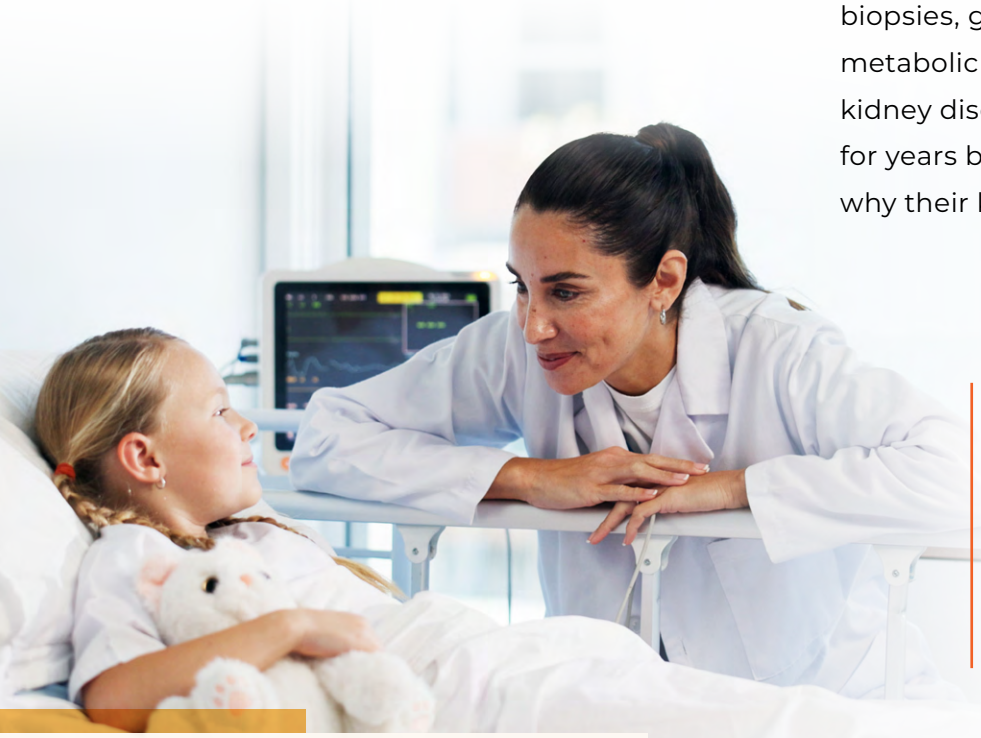
Rare kidney diseases disproportionately affect children and young adults. In fact, most kidney diseases in children are rare kidney diseases, and they account for more than half of kidney failures among children.

The consequences extend well beyond physical health. Young patients with rare kidney disease often experience anxiety and depression, and many miss school, work and time with friends — setbacks that can shape the rest of their lives. As their health declines, so does their independence, and the strain on parents and family members grows. Families face missed paychecks on top of the costs of dialysis trips and transplant surgery.

Q: Why are rare kidney diseases often misdiagnosed?

Early symptoms of kidney disease are easy to miss. By the time most patients are diagnosed, half their kidney function is already gone. The low prevalence of rare kidney conditions makes diagnosis and treatment challenging, but the impact of misdiagnosis is significant. Standard kidney screening — urine and blood tests — can identify a decline in kidney function, but not what's causing it. As a result, many people with a rare kidney disease are misdiagnosed with the more common chronic kidney disease, often resulting from unmanaged high blood pressure or diabetes.

Identifying the actual cause of declining kidney function takes more work: kidney biopsies, genetic tests, imaging or tests for metabolic problems. A person with a rare kidney disease may bounce between providers for years before getting a clear answer as to why their kidney function is in decline.



Nearly 58% of children on kidney replacement therapy have a rare kidney disease.[6]

Q: Why is identifying the cause of rare kidney disease so important?

Identifying the cause of declining kidney function can change a patient's life. With the correct diagnosis, doctors can prescribe treatments aimed at the specific disease.

For people at risk for a rare kidney disease, particularly young people experiencing declining kidney function, identifying the cause also opens the door to clinical trials and, for families with genetic conditions, to receive genetic counseling and family planning.

> Knowing what's causing kidney decline opens the door to targeted treatment and life-changing outcomes.

Q: What treatments are currently available for rare kidney diseases?

For people experiencing chronic kidney disease, general treatments such as lifestyle changes and medications can help slow decline. But for a growing list of rare kidney diseases, there are now medications aimed at the specific disease itself. More medications are in development, and gene-based therapies are advancing in clinical trials.

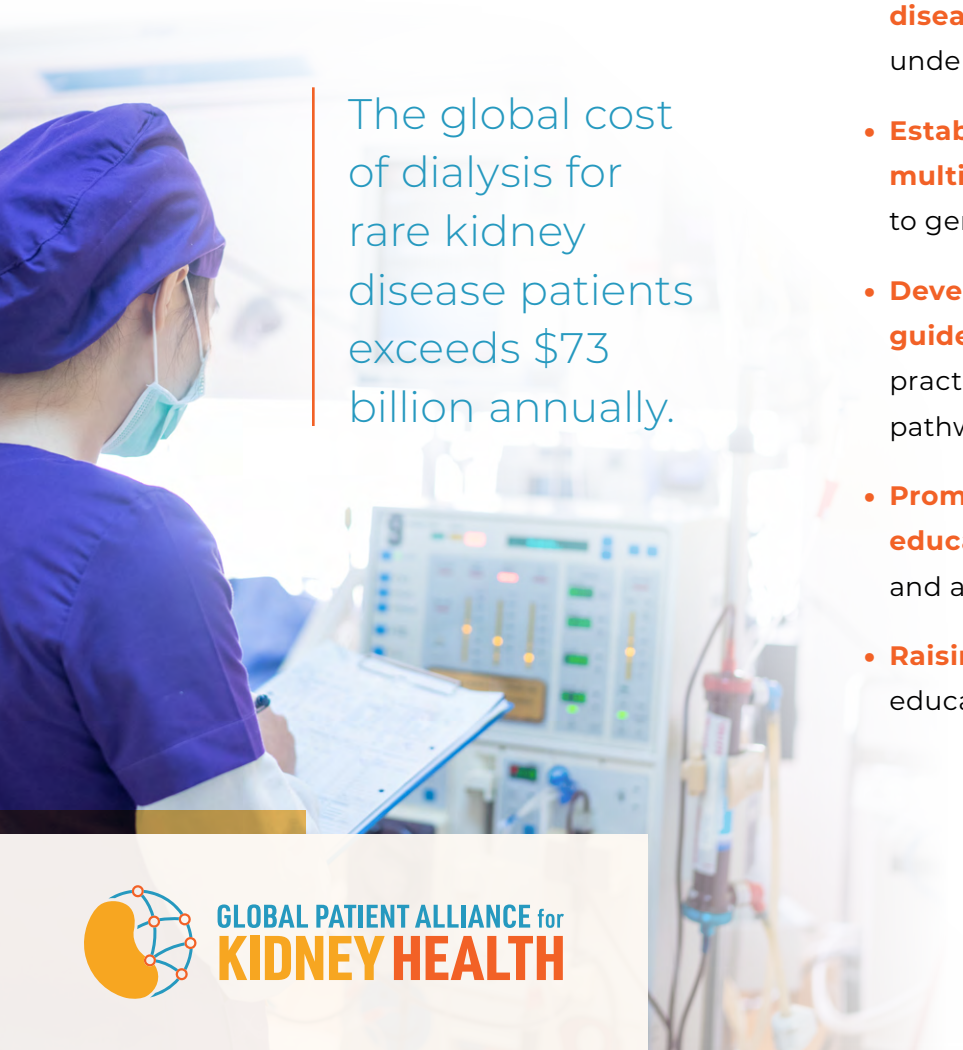
These targeted treatments can preserve kidney function and meaningfully delay — or even prevent — kidney failure. But patients can only access them if their disease has been correctly identified.

> New treatments are offering new promise for some rare kidney diseases.

Q: Who else benefits from accurate diagnosis of rare kidney diseases?

Beyond the person treated for a rare kidney condition, the benefits extend to caregivers. When patients stay healthier longer, families spend less time driving to appointments and managing care.

The stakes for health systems are just as high. Advanced kidney disease drives more hospital visits and higher drug costs. Dialysis alone for rare kidney disease patients costs more than $73 billion globally each year.[7] Dialysis also takes a real environmental toll — large amounts of freshwater use, single-use plastic and energy.



> The global cost of dialysis for rare kidney disease patients exceeds $73 billion annually.

Q: How can policymakers and health systems support early, accurate diagnosis of rare kidney diseases?

The burden of rare kidney diseases is significant — and so is the opportunity for meaningful change. Policymakers and health systems can prioritize identifying the cause and treatment of rare kidney diseases by:

- **Increasing access to screening** for declines in kidney function
- **Prioritizing the earlier diagnosis and treatment** of rare and chronic kidney diseases
- **Supporting international rare kidney disease research** to improve disease understanding
- **Establishing centers of excellence and multidisciplinary teams** to provide access to genetic and other advanced tests
- **Developing and implementing clinical guidelines** to help standardize best-practices care and disease-specific pathways based on current evidence
- **Promoting health care provider education** so they conduct the right tests and an accurate diagnosis
- **Raising public awareness** through education programs

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CONCLUSION

Rare kidney diseases may affect a small share of the population individually, but together they represent significant and under-addressed drivers of kidney failure worldwide. Their impact falls hardest on children, young adults and the caregivers who support them.

But early, accurate diagnosis and access to targeted treatments can dramatically improve outcomes. Advances in genetics, immunology and drug development are redefining what's possible. Now, policies that expand access to diagnostics and innovative treatments can relieve the growing burden on patients and health systems.

To learn more about the importance of understanding the cause of rare kidney diseases and how policy solutions can support, read GloPAKH's policy brief **Outsized Impact: Why Identifying the Cause of Rare Kidney Disease Matters.**

References

1. Jager KJ, Kovesdy C, Langham R, Rosenberg M, Jha V, Zoccali C. A single number for advocacy and communication-worldwide more than 850 million individuals have kidney diseases. Nephrol Dial Transplant. 2019;34(11):1803-5.
2. Global Burden of Disease Chronic Kidney Disease Collaboration. Global, regional, and national burden of chronic kidney disease, 1990-2017: a systematic analysis for the Global Burden of Disease Study 2017. Lancet. 2020;395(10225):709-33.
3. European Commission. Rare diseases. Available at: https://health.ec.europa. eu/rare-diseases-and-european-reference-networks/rare-diseases_ en#:~:text=In%20the%20EU%2C%20rare%20diseases,rare%20diseases%20 start%20in%20childhood.
4. Connaughton DM, Kennedy C, Shril S, Mann N, Murray SL, Williams PA, et al. Monogenic causes of chronic kidney disease in adults. Kidney Int. 2019;95(4):914-28
5. Jefferis J, Hudson R, Lacaze P, Bakshi A, Hawley C, Patel C, Mallett A. Monogenic and polygenic concepts in chronic kidney disease (CKD). J Nephrol. 2024;37(1):7 21.
6. Wuhl E, van Stralen KJ, Wanner C, Ariceta G, Heaf JG, Bjerre AK, et al. Renal replacement therapy for rare diseases affecting the kidney: an analysis of the ERA-EDTA Registry. Nephrol Dial Transplant. 2014;29 Suppl 4:iv1-8.
7. Vanholder R, Coppo R, Bos WJW, Damato E, Fakhouri F, Humphreys A, et al. A Policy Call to Address Rare Kidney Disease in Health Care Plans. Clin J Am Soc Nephrol. 2023;18(11):1510-8.

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Alexion has provided a financial sponsorship to the Global Alliance for Patient Access as the secretariat of the Global Patient Alliance for Kidney Health.