Collectively common: the devastating impact of rare kidney diseases in the UK

A call for investment, innovation and quality care

September 2025

Kidney Research UK

ZS

Disclaimer

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Tables and figures

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About this report

Kidney Research UK commissioned ZS Associates to prepare an independent report on the impact of rare kidney diseases in the UK. The report provides an overview of the evolving clinical, regulatory and policy landscape of rare kidney diseases in the UK; the impact on patients, families and the broader economy, illustrated by personal stories from people affected by rare kidney diseases; and strategic policy, research and clinical recommendations to ultimately improve rare kidney disease care and outcomes in the UK.

This report was prepared by ZS Associates in collaboration with Kidney Research UK and the expert advisory steering group in 2025.

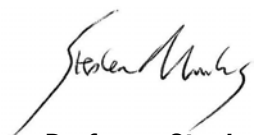
Endorsement

The UK Kidney Association (UKKA) is pleased to endorse this report, informed by extensive clinical expertise from our colleagues. We have confidence in the rigour of the process and high-quality clinical input, which ensures that these findings are robust and actionable. This report is an important, evidence-based contribution to our collective efforts in understanding and improving management of rare kidney diseases across all ages.

As the leading professional body for the UK kidney community, the UKKA is dedicated to improving lives by supporting professionals involved in the delivery of kidney care and research. We welcome initiatives that equip clinicians with the knowledge, tools and data they need to drive improvement in care for people with rare kidney diseases. We look forward to disseminating and implementing the report's recommendations across the professional kidney community.

Under the UKKA umbrella, the British Association of Paediatric Nephrology (BAPN) and Renal Pharmacy Group, also warmly welcome this report, which specifically highlights inequities in research and access to novel treatments for children with rare kidney diseases. We too look forward to sharing and implementing its findings around new medications and seeking opportunities to bridging the gap between paediatric and adult services.

Claire Morlidge
Joint President, UK Kidney Association



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About Kidney Research UK

Kidney Research UK is the leading charity in the UK focused on funding research into the prevention, treatment and management of kidney disease.

Our vision is the day when everyone lives free from kidney disease, and for more than 60 years the research we fund has been making an impact.

But kidney disease is increasing, as are the factors contributing to it, such as diabetes, cardiovascular disease and obesity, making our work more essential than ever.

At Kidney Research UK we work with clinicians and scientists across the UK, funding and facilitating research into all areas of kidney disease. We collaborate with partners across the public, private and third sectors to prevent kidney disease and drive innovation to transform treatments.

Over the last ten years we have invested more than £71 million into research.

We lobby governments and decision makers to change policy and practice to ensure that more than seven million people living with kidney disease in the UK have access to the most effective care and treatment, and to make kidney disease a priority.

Most importantly, we also work closely with patients, ensuring their voice is heard and is at the centre of everything we do, from deciding which research to invest in to how we plan our priorities and our work across the charity.

Those patient contributions are vital, always helping us and our partners to understand what life is like with kidney disease, always ensuring we see the patient behind the treatment and always reminding us that behind every statistic and every number are people – the patients and the carers who inspire our mission and push us forward to make a difference and change the future of kidney disease.

Foreword

Dame June Raine DBE
Former Chief Executive,
Medicines and Healthcare Regulatory Authority (MHRA)

This timely report comes at a pivotal moment for people with kidney disease. We are entering an era where understanding the molecular basis of an individual patient's disease is increasingly possible, supporting earlier diagnosis. For people with rare kidney diseases, this offers immense opportunities for targeted therapies and personalised care.

This report sets out a powerful case for change by shining a light on the breadth and complexities of rare kidney diseases, mixing research and evidence alongside patient experiences and clinical insight. For those living with one of these diseases, the impact is profound. They may represent between just 5% and 10% of people living with the most advanced stages of kidney disease, but when it comes to kidney failure, they make up 25% of the adult patient population and 60% of the paediatric population, enduring treatments that seriously impact their life chances.

In 2023, Kidney Research UK published a major report which painted a stark picture of the current reality and future picture of kidney disease, which is forecast to be the fifth leading cause of premature death globally by 2040. The economic burden in the UK was projected to grow from £7 billion to £13.9 billion by 2033, whilst demand for dialysis could overwhelm the NHS.

I welcome this new report as a call to action for rare kidney disease. This patient group remains significantly underserved. Diagnosis is delayed. Research and innovation are increasing, and although there is a burgeoning pipeline of exciting new drugs, new treatments remain years away. In particular, the current approach to drug development means access to new medicines for children lags behind adults by up to 13 years. Most patients rely on decades-old treatments that manage symptoms but are not a cure, whilst unequal care pathways undermine outcomes.

If taken forward, the clear recommendations in this report could transform care for people with rare kidney diseases. But closing the gaps in treatment access, delays in paediatric access to therapies, poor transition from paediatric to adult services, and underinvestment in research will require sustained commitment, cross-sector collaboration and bold policy action. The time to make that commitment and take that action is now.

June M. Raine

Dame June Raine DBE

Context

Kidney disease is a public health emergency costing the UK economy £7 billion annually, with rare kidney diseases – a group of over 150 distinct conditions – contributing disproportionately. Although individually affecting fewer than 1 in 2,000 people, we estimate that these rare diseases affect over 160,000 people in the UK.

People with rare kidney diseases tend to reach kidney failure at a younger age and spend more of their lives needing treatment than those with more common causes of chronic kidney disease (CKD). Consequently, they require treatment such as dialysis for longer leading to markedly higher lifetime costs. Despite representing less than 10% of the CKD population, people with rare kidney diseases disproportionately account for more than 25% of dialysis and transplant patients. It is also important to note that dialysis and transplants are not curative: people endure disrupted education, employment, and family life, and the NHS bears significant long-term costs.

In May 2025, the World Health Assembly prioritised kidney and rare diseases, catalysing research momentum. Promising therapies are entering trials, but most remain years from approval. The effects on children with rare kidney diseases are compounded due to exclusion from research, with access to new medicines lagging behind adults for up to 13 years.

This report calls for urgent action to transform care. Key priorities include:

- raising awareness and policy focus
- improving diagnostics through biomarkers and genetic testing
- accelerating evidence generation and clinical trial access
- streamlining access to new medicines; and
- personalising care to reflect disease heterogeneity.

Despite some progress, the rare kidney disease community remains underserved. Bridging gaps in treatment, diagnosis, and care transitions demands sustained investment, cross-sector collaboration, and bold policy reform – ensuring that innovation and quality care reaches every patient, regardless of how rare their condition is or where they live.

Key findings

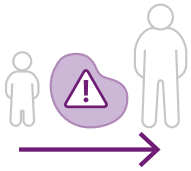
1 **Rare kidney diseases include over 150 distinct diseases** that, while individually uncommon, are estimated to collectively affect around **160,000** people in the UK

2 We estimate that rare kidney diseases cause between 5% and 10% of all cases of CKD at the most advanced stages but disproportionately account for **25% of all dialysis and transplant patients**

3 The cost to the NHS of dialysis for rare kidney diseases **is approximately £263 million per year**

4 Rare kidney diseases disproportionately affect children: **more than 60% of children with kidney failure have a rare kidney disease**

5 Children with rare kidney diseases are **likely to be on dialysis or living with a kidney transplant by the time they reach adulthood**



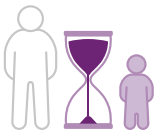
6 Children on dialysis will on average **miss more than 100 days of school per year**, while one person interviewed for this report **missed an entire year of school**

7 In addition to having worse health, **people with rare kidney diseases are economically disadvantaged**: the majority of people with a rare kidney disease have a **64% chance of being out of work by the time they reach kidney failure**

8 Despite the significant impact of rare kidney diseases on economic productivity, this is underresearched in the UK: a global systematic review identified 33 productivity studies on the impact of kidney disease on patients' ability to work, but **none covered the UK**

9 There are a limited number of new targeted therapies in development for several rare kidney diseases; however, of the 13 we reviewed, **about 40% of trials did not include a UK centre**, meaning UK patients do not get the opportunity to try new therapies soon enough, and the UK economy is missing out on R&D investment

10 The impact on children is compounded as they are almost universally excluded from trials: as a result, **treatment innovations for children lag by up to 13 years** behind those for adults



Executive summary

Rare kidney diseases include over 150 distinct diseases that, while individually uncommon, collectively affect around 160,000 people in the UK. They account for 5%–10% of CKD cases at the most advanced stages. These lifelong diseases can lead to kidney failure requiring dialysis or a kidney transplant and have a heavy and often hidden impact on families and the NHS. Despite this, rare kidney diseases remain underrecognised in health policy, research funding and clinical services. Delays in diagnosis, limited effective treatments and fragmented care systems lead to suffering that could be avoided and to significant costs, both personal and economic.

Nearly 80% of these conditions are inherited, and many begin in childhood. Most children who develop kidney failure do so because of a rare kidney disease. Over 25% of adults and more than 60% of children receiving dialysis or a kidney transplant are affected by a rare kidney disease. Although many types begin in early life, the average age at diagnosis across the UK National Registry of Rare Kidney Diseases (RaDaR) is 41, highlighting how long some people wait for a correct diagnosis. These delays cause physical and emotional harm and prevent timely access to specialist care. The purpose of this report is to raise awareness of the impact of rare kidney diseases in the UK and to call for action to improve outcomes for every person affected. It provides an overview of rare kidney diseases in the UK, including:

- Epidemiology and heterogeneity of rare kidney diseases
- Impact on patients, the NHS and the economy
- Current policy landscape
- Potential for innovations and recommendations for bringing them to patients more swiftly

Research methods utilised included:

- Targeted review of academic literature
- Further pragmatic search of academic and grey literature
- Expert input from leading UK clinical experts
- Case studies from nine patients or their carers across four rare kidney diseases

Summary and recommendations

Diagnostic challenges

Rare kidney diseases may mimic common conditions and are often asymptomatic at early stages, leading to delayed diagnoses or misdiagnoses. Delays in diagnosis can lead to further disease progression, and misdiagnoses can lead to inappropriate treatment, leaving **patients with worse outcomes and closer to kidney failure**.

Recommendation 1: Ensure sufficient and equitable access to diagnostics, including genetic testing

Recommendation 2: Develop targeted screening programmes for people at known risk of rare kidney diseases

Recommendation 3: Enhance education and support for kidney specialists and primary care providers to improve early recognition of potential rare kidney diseases and ensure timely, appropriate referrals for further evaluation and diagnosis

Healthcare challenges

NHS care pathways are not well adapted to the needs of people with rare kidney diseases, particularly children. Renal clinical networks and paediatric renal services improve coordination but lack national consistency, and there is inadequate support for transition from paediatric to adult care. In addition, regional access to specialist care is inconsistent. **Rare kidney disease care should be prioritised as the UK Government develops new policy to replace the current UK Rare Diseases Framework and ensure it is implemented across the four nations, to manage the disproportionate impact of rare kidney diseases.**

Recommendation 4: Strengthen interconnection and collaboration between specialist centres and other health settings, e.g. via renal networks

Recommendation 5: Improve integration and continuity of care between paediatric and adult services, including increased support for transitional care and adolescent mental health services, and secure additional training for adult nephrologists on paediatric kidney diseases

> As more children with rare kidney diseases are now surviving into adulthood, nephrologists treating adults are increasingly encountering conditions they may not have seen before. Historically, many of these were considered childhood diseases. To keep pace with this shift, adult nephrologists need additional training and support to recognise and manage rare kidney diseases that may be unfamiliar to them."
>
> **Dr Ben Reynolds, Consultant Paediatric Nephrologist**

Access to new medicines

The limited availability of disease-specific treatments means many people with rare kidney diseases rely on supportive or generalised treatments with considerable side effects. There is some hope, however, with a number of clinical trials ongoing for new targeted medicines. It is essential that regulatory and reimbursement pathways for medicines are optimised for rare diseases to **ensure patients receive access to new treatments as soon as possible**:

Recommendation 6: Ensure coordination between regulatory and reimbursement evidence needs so that rare disease studies are appropriately designed and inclusive across age groups

Recommendation 7: Include people with rare kidney diseases in research studies and clinical trials so that future treatments for mixed causes of CKD are also understood in rare diseases

> Gareth's kidneys were slowly deteriorating, but we embraced life together… We held on to the thought that advances in medicine could bring better therapies, or even a cure, in his lifetime." Due to a lack of treatment options for his autosomal dominant polycystic kidney disease (ADPKD) beyond hypertension control, he required heart and kidney transplants. He died aged 50.
>
> **Caroline Prodger, wife of Gareth**

Clinical trials

Enrolment in a clinical trial offers participants early access to new medicines and can boost UK R&D investment. However, in a recent review of 13 clinical trials for rare kidney diseases, 40% did not have a UK site, **meaning UK patients and the UK economy are not benefiting as much as they could be**. Furthermore, the exclusion of children from rare kidney disease trials means they are unable to benefit from new therapies at the same time as adults.

Recommendation 8: Support UK participation in regional and international clinical trials to increase UK patient access and representation

Recommendation 9: Address inequities around the exclusion of children from clinical trials where this is safe and appropriate

Recommendation 10: Offer everyone with a rare kidney disease the opportunity to participate in a clinical trial or research for their disease

Enhance RaDaR

RaDaR is a world-leading patient registry and an invaluable resource for the study of rare kidney diseases in the UK. However, enrolment into the database is time-consuming, and there is no government funding to support patients wishing to do so. This means **we lack an accurate picture of the true prevalences of rare kidney diseases in the UK, and the data we do have are geographically skewed. Furthermore, patients who might benefit from participating in research are missing out**.

Recommendation 11: Increase capacity to enrol patients earlier, growing the cohort over time, to facilitate and expedite recruitment for clinical trials

Recommendation 12: Increase communication and education within the rare kidney disease community in the UK to foster trust and information exchange between patients and clinicians, and within RaDaR and beyond

Kathryn Croker was 13 when she was diagnosed with IgA vasculitis. Within five months, Kathryn's kidneys failed. She missed an entire year of school and, now in her twenties, works reduced hours due to ongoing symptoms and extreme tiredness.

Lack of economic evidence

Primary research into the economic impact of rare kidney diseases is lacking. A systematic review of the economic literature focused on rare kidney diseases published over the last ten years found 33 studies globally, but **none covered the UK**.

Recommendation 13: Demonstrate the economic value of delaying the progression of rare kidney diseases — either by reporting these conditions separately in large studies or conducting specific ones, e.g. through expansion and provision of access to the underlying dataset to researchers of the 2025 Office for National Statistics report: impact of health conditions requiring hospitalisation on earnings

Recommendation 14: Ensure rare kidney diseases are accurately captured in administrative data, which inform cost analyses

Rare kidney diseases are a complex and underaddressed challenge within the UK healthcare system. While individually uncommon, their collective burden on patients, families and the NHS is substantial, ranging from delayed diagnosis and fragmented care to limited treatment options and minimal economic data. Patient experiences throughout this report highlight the personal and systemic toll of these conditions. To meaningfully improve outcomes, a coordinated response is needed that combines earlier detection, integrated and equitable care pathways, investment in targeted therapies and clinical research, and improved national data infrastructure. Implementing the recommendations outlined here is critical to ensuring that people with rare kidney diseases are no longer overlooked in policy, research and care delivery.

Introduction

The kidneys play a vital role in maintaining overall health by filtering waste and excess fluid from the blood to produce urine. Each kidney contains approximately one million tiny filtering units called nephrons, which remove waste while returning essential substances, such as water, salts and minerals, to the bloodstream. In doing so, the kidneys help regulate blood pressure, support the production of red blood cells, produce hormones and maintain bone strength.[1,2] Kidney disease occurs when the kidneys are damaged and lose their ability to filter blood effectively. It is a serious health condition that can lead to kidney failure – the end point of disease progression – where a person needs either dialysis or a kidney transplant to survive.[1,3] A distinct subset of kidney diseases are the rare kidney diseases, encompassing at least 150 different conditions, most of which have an onset in childhood or young adulthood.[4–6]

> My symptoms never felt serious or interconnected… I was diagnosed with IgA nephropathy [and] was told my kidney function had fallen to 25%. At every subsequent hospital visit, it dropped further, until my kidneys began to fail. It was a rapid decline, and the need for a kidney transplant became urgent to avoid dialysis."
>
> **Paul Vallois, 49-year-old with IgAN**

Overview of kidney disease

Kidney disease encompasses a broad range of conditions that affect people of all ages, genders, and ethnicities.[1,7,8] There is currently no cure for kidney disease, so treatment is focused on delaying disease progression and managing complications.

Frequently described as a silent killer, kidney disease often progresses without noticeable symptoms.[1,7,8] Chronic kidney disease (CKD) is defined as abnormalities in the structure or function of the kidneys that have been present for at least three months, impacting overall health. CKD affects an estimated 7.2 million people in the UK – more than 10% of the population – and its prevalence is increasing due to an ageing population and rising rates of known risk factors such as diabetes, high blood pressure and cardiovascular disease.[6,9,10] Most rare kidney diseases lead to CKD and account for 5%–10% of CKD cases in adults.[6,9,10] We estimate there are 160,000 people with rare kidney diseases in the UK, around 5% of the 3.25 million people living with CKD stages 3–5.[6,10] Early detection of CKD, including some rare kidney diseases, remains low due to the lack of early symptoms and limited screening programmes.

CKD progresses through five stages (see Figure 1), which also apply to rare kidney diseases, each defined by the level of kidney function remaining.[11] In stages 1–2, there is evidence of kidney damage and mild loss of function, yet most people remain unaware of any problems. Stage 3 marks a more noticeable decline and is often when kidney disease is diagnosed, though many people still experience no symptoms. By stage 4, kidney function is severely reduced, and by stage 5, kidney failure has occurred.[11] At this point, survival requires significant medical intervention to replace kidney function in the form of dialysis or kidney transplant, while some patients may choose to focus on symptom management and quality of life through supportive care.

Figure 1. Stages of chronic kidney disease.

Stages of chronic kidney disease	Percentage of kidney function	Symptom/implication
STAGE 1 Kidney damage with **normal** kidney function	100–90% 	People in early-stage CKD may not know they have CKD as they often feel well and show no symptoms
STAGE 2 Kidney damage with **mild loss** of kidney function	89–60% 	
STAGE 3a **Mild to moderate** loss of kidney function	59–45% 	People are often diagnosed with kidney disease in the mid-stage, with many people still asymptomatic as waste in the body builds and blood pressure rises
STAGE 3b **Moderate to severe** loss of kidney function	44–30% 	
STAGE 4 **Severe** loss of kidney function	29–15% 	
STAGE 5 Kidney **failure**	Less than 15% 	Patients with kidney failure require dialysis (a type of kidney replacement therapy that replaces the blood-filtering function of the kidneys) or a kidney transplant to stay alive A proportion of people with kidney failure will not receive either dialysis or transplant, instead undergoing supportive care

The average wait for a kidney transplant is currently two to three years for adults and around two years for children, with approximately 7,000 people on the waiting list.[12,13] In 2024–2025, more than 3,000 kidney transplants were performed in the UK.[13] Overall, the number of donor kidneys available is not keeping up with demand, leading to prolonged wait times and preventable morbidity and mortality. Transplants from deceased organ donors typically last an average of 15–20 years, while those from living donors last 20–25 years.[13] Individual outcomes vary depending on factors like age, health status and coexisting conditions such as diabetes and heart disease.[13] Pre-emptive transplantation – where a kidney transplant is performed before dialysis is needed – offers the best outcomes for patients but remains underutilised in both children and adults due to challenges in early diagnosis, donor availability and care coordination.[14]

Beyond clinical risks, environmental and social factors – such as social inequalities and genetic predisposition – also play a role in the development of kidney disease. Health disparities disproportionately affect those from lower socioeconomic backgrounds, and some minority ethnic groups, who are more likely to develop CKD, experience faster progression and have poorer outcomes. For example, in CKD populations, some minority ethnic groups are three to five times more likely to require dialysis and face significantly longer waits for kidney transplants.[8,10,15] The effects of social and environmental factors may be even more pronounced for rare kidney diseases.[4,16]

While dialysis and kidney transplant offer life-sustaining support for people with kidney failure, early detection and intervention remain critical to improving outcomes.[3,11]

> Kidney disease made me feel sad, as I was always poorly and spent a lot of time in hospital. My dialysis machine put warm water in my tummy to help stop me getting poorly. George, my new kidney, makes me happy and healthy and gives me energy to do stuff."
>
> **Poppy Lancaster, seven-year-old transplant recipient with polycystic kidney disease**

Economic burden of kidney disease

Based on Kidney Research UK's 2023 report into the economic impact of kidney disease, the total cost of kidney disease to the UK economy was estimated at £7.0 billion per annum.[10] This includes £6.4 billion in direct costs to the NHS, approximately 3.2% of the £197 billion of total NHS spending across the four nations.[10] The £7.0 billion estimate also includes £372 million in productivity loss for people receiving dialysis and those who support them. A further £225 million is spent on transport costs for people to and from dialysis centres.[10]

Kidney failure accounts for a disproportionately large share of this total. On average, in 2022, hospital haemodialysis costs about £34,000 per year per patient (£38,670 in 2025 adjusting for inflation), with travel to and from dialysis adding up to £8,000 (£9,100) annually, while a kidney transplant costs £18,145 (£20,640).[17] Since people with rare kidney diseases are more likely to reach kidney failure compared to those with more common kidney diseases, rare kidney diseases account for a disproportionate share of the total cost of kidney care.

Aims of this report

Our previous report, 'Kidney Disease: A UK Public Health Emergency' (2023),[10] focused on the overall impact of kidney disease on the UK health system and economy. This new report analyses the impact rare kidney diseases have on that landscape and highlights the personal experiences of those directly affected.

In this report, we conceptually organise and contextualise the current state of knowledge and care for rare kidney diseases by outlining the broad spectrum of conditions, their epidemiology, and the substantial burden they impose on patients, caregivers, the NHS and the economy. Given the heterogeneous nature of these rare diseases, the report considers them collectively but also provides four in-depth case studies of people with specific diseases. These focused analyses highlight some key themes and challenges and provide targeted recommendations for enhancing care and outcomes.

Rare kidney diseases

Rare kidney diseases are underdiagnosed and often misunderstood. This gap in recognition among clinicians, the public and policymakers delays diagnosis, hinders access to treatment (if available) and limits the adoption of new advances in care. Added to these challenges are the high costs of developing and bringing new treatments to market, as well as the difficulty of conducting scientifically rigorous clinical trials in small patient populations, which further complicate efforts to deliver timely therapies for rare conditions. In addition, smaller patient populations further disincentivise industry to invest in developing new treatments.

Overview of rare kidney diseases

Rare kidney disease is defined in the UK and the European Union as a kidney condition affecting fewer than 1 in 2,000 people. Current estimates indicate that over 35,000 people in the UK are affected by serious kidney impairment attributable to more than 150 distinct rare kidney diseases, many of which are genetic or autoimmune in nature and disproportionately affect children.[6]

Rare kidney diseases are also over-represented among individuals requiring dialysis and kidney transplants. While rare kidney diseases affect 5%–10% of the general kidney disease population, they account for more than 25% of all patients receiving dialysis and kidney transplants and more than 50% of paediatric kidney transplant recipients.[6,18] In people with kidney failure, over 25% of adults and 60% of children have an underlying rare kidney disease.[6,18,19] This disproportionate representation reflects the early onset, progressive nature and often limited treatment options available for rare kidney diseases.[6,18]

Beyond direct healthcare costs, the indirect economic and social impact of rare kidney diseases remain poorly understood. Families often face challenges such as lost income, educational disruption, long-term caregiving responsibilities and emotional strain – yet these effects have not been systematically studied in the UK. This represents a major gap in the evidence base and a barrier to fully understanding the impact of rare kidney diseases on patients, families and society. These personal daily challenges are highlighted in the patient stories that appear throughout this report.

"[My husband] Ian and I don't get to see each other much because one of us always needs to stay in hospital with Asher, and we both need time at home with our other children. I own a hair salon but had to step away from my job because I'm never at home. We have been financially affected, but Ian continues to work full time to provide for us… we've been lucky that my mum has been able to move in with us to look after our other children while we are not home."

Eloise Pyper, mother of two-year-old Asher, who was born with posterior urethral valves, a congenital anomaly of the kidneys and urinary tract (CAKUT)

The rarity and heterogeneity of these diseases also pose challenges for timely diagnosis, coordinated care and equitable access to specialist services.[5,9,19] In addition, for many rare kidney diseases, there are no specific approved treatments available, leaving people with kidney diseases reliant on symptom management or non-targeted therapies, further compounding the burden and limiting opportunities for improved outcomes.[20,21] Even where treatments do exist, there remains a critical need for more effective, better-tolerated and disease-modifying medications.

Based on the significant impact demonstrated, rare kidney diseases warrant targeted policy attention, investment in research and tailored models of care to improve outcomes and optimise resource allocation across the healthcare system.[5,9,19]

Challenges in the management of rare kidney diseases

Accurately diagnosing rare kidney diseases remains one of the most pressing and persistent challenges. Many of these diseases present with non-specific symptoms or resemble more common renal or systemic diseases, leading to frequent misdiagnoses, diagnostic delays or ineffective treatments.[5] These challenges are especially acute in children, as medicine is mainly oriented around adults with common conditions. Care for children with rare kidney diseases suffers due to limited investment, a shortage of academic paediatric nephrologists and regulatory constraints that often require treatments to be tested in adults first, delaying access to new therapies for younger patients. This prolonged uncertainty can substantially postpone the initiation of appropriate treatment, if available, increasing the risk of irreversible kidney damage and accelerating progression to kidney failure.[5]

> I try to live my life to the fullest despite the setbacks, particularly for my wife and two children who I am blessed with. It's frustrating to know that if my kidney disease had been detected sooner, I might not have gotten so ill, so it is my hope that new patients are diagnosed much sooner."
>
> **Adam Musa, born with congenital renal dysplasia which remained undetected until he was diagnosed at 21 years old**

People with rare kidney diseases typically have a limited range of therapeutic options.[22] For many disorders, there are no disease-specific treatments available, and clinical management remains largely generic. Interventions are often based on generalisation from treatments used in more prevalent conditions, which do not adequately address the underlying disease mechanisms.[20,23] This scarcity of targeted therapies reflects the broader challenges inherent in developing new treatments for rare diseases.[23]

Several systemic and scientific barriers hinder the development of novel therapies. These include limited understanding of disease pathophysiology due to small patient numbers, a lack of agreed clinical endpoints for clinical trials, and insufficient natural history data.[16,20,23] Fragmented care pathways and variability in diagnostic coding further obscure disease tracking and impede recruitment for research.

In children with rare kidney diseases, collecting and storing biological samples (biobanking) presents additional challenges. Parental permission and the child's agreement are usually needed, and stricter ethical rules apply.[7] Only small biological samples can be taken – especially from younger or seriously ill children – and it is often difficult to track children over time as they move between services.[7] Without dedicated investment in child-friendly research infrastructure, valuable opportunities to improve diagnosis and treatment for childhood-onset rare kidney diseases will be missed.[7]

As a result, there have been relatively few clinical studies and randomised controlled trials focusing on rare kidney diseases for both paediatric and adult patients.[24] The small and geographically dispersed patient population makes trial recruitment difficult, particularly in earlier stages of disease when individuals may remain undiagnosed. Trials in children face additional barriers and, in many cases, fail to recruit sufficiently or terminate early due to feasibility challenges, as recently highlighted.[25] In addition, the complexity of trial design – including the need to define meaningful, disease-specific outcomes – and the demands of regulatory approval processes present additional hurdles. Together, these factors contribute to a persistent gap in the evidence base, slowing innovation and limiting the development of, and access to, new treatments for those affected.[5,20,23]

These challenges create a situation in which people living with rare kidney diseases face considerable uncertainty, not only in receiving a timely diagnosis and appropriate treatment but also in accessing emerging therapies and participating in research. This uncertainty is further compounded by the wide variation in how these diseases present and progress; even individuals with the same diagnosis may experience vastly different outcomes, and many do not know whether or when they will require dialysis or a kidney transplant.

Addressing these issues requires coordinated investment in rare kidney disease infrastructure, enhanced data and biological sample collection, and targeted incentives to drive innovation in this underserved area of kidney health.[5,20,23]

Classifying rare kidney diseases

Rare kidney diseases can be congenital, genetic or autoimmune in origin.[26,27] Data from the UK's National Registry of Rare Kidney Diseases (RaDaR) provide important insights into the distribution, clinical outcomes and treatment needs of these diseases across different age groups (Appendix Table 1).[6,18]

Rare kidney diseases demonstrate striking heterogeneity in prevalence, age of onset and clinical progression. UK patient numbers range from under 100 in ultra-rare conditions such as HNF1B mutations and hyperoxaluria to several thousand in other disorders like autosomal dominant polycystic kidney disease (ADPKD) and IgA nephropathy (IgAN).[18] The median age at diagnosis spans from infancy in conditions such as congenital anomalies of the kidney and urinary tract (CAKUT; the most common cause of kidney failure in young children) to late adulthood in diseases such as monoclonal gammopathy of renal significance (MGRS, 63 years).[6] Similarly, disease progression varies substantially – while diseases like membranous nephropathy and MGRS are associated with a relatively rapid decline in kidney function, others such as tuberous sclerosis complex or minimal change nephrotic syndrome (MCNS), show a much slower trajectory.[6]

Mortality patterns also diverge, with some diseases showing extended survival post-diagnosis and others associated with early mortality.[6] For example, cardiovascular mortality is significantly higher in inflammatory kidney diseases such as anti-neutrophil cytoplasmic antibody (ANCA)-associated vasculitis, likely due to the long-term effects of persistent systemic inflammation.[28–30] This variation reinforces the importance of disease-specific approaches to research, service planning and patient management. It also increases the complexity of developing and evaluating treatments, as therapies must often be tested at different stages of CKD – adding further challenges to trial design, regulatory approval and achieving commercial viability.

A substantial proportion of rare kidney diseases are genetic or inherited disorders that result from variants in genes essential for normal kidney development or function.[5,19] For example, cystinosis is caused by defective variants in the CTNS gene, leading to the accumulation of cystine within cells and resulting in Fanconi syndrome (where kidneys lose essential nutrients like sugar, salts and proteins in the urine), growth failure and progressive kidney dysfunction. Alport syndrome, a collagen-related disorder, typically presenting in childhood, is caused by variants in the COL4A3, COL4A4 or COL4A5 gene and leads to progressive kidney decline, often with hearing loss and eye problems that can emerge later. ADPKD, caused by mutations in the PKD1 or PKD2 gene, is more prevalent than most rare kidney diseases and is a major contributor to kidney failure in adults, diagnosed by the development of fluid-filled kidney cysts.[5,19] Its rarer counterpart, autosomal recessive polycystic kidney disease (ARPKD), presents in childhood, often with severe kidney and liver involvement, and is associated with mutations in the PKHD1 gene. Many affected children require both kidney and liver transplants, and some die in infancy or early childhood, meaning the true burden of the disease may be underreported.[5,19]

Autoimmune-mediated kidney diseases, though less common in children, are a major group of rare kidney diseases in adults.[5,19] These are disorders where the immune system erroneously attacks kidney tissue, causing inflammation and progressive damage. ANCA-associated vasculitis with renal involvement damages the kidney's filters very quickly so they are unable to clean the blood properly, and it can lead to kidney failure if not appropriately managed.[31] Granulomatosis and anti-glomerular basement membrane disease are other key examples, both capable of causing rapidly deteriorating kidney function and, in the case of the latter, causing life-threatening pulmonary haemorrhage.[5,19] These conditions require timely diagnosis and immunosuppressive treatment to prevent irreversible kidney damage.

Despite their heterogeneity – from structural anomalies to genetic and autoimmune disorders – rare kidney diseases share common challenges: delayed diagnosis, limited treatment options and poor long-term outcomes. The RaDaR dataset highlights just how varied these conditions are in onset, progression and mortality, reinforcing the need for tailored and coordinated approaches to care and research. As the UK continues to invest in rare disease infrastructure and innovation, it remains crucial to prioritise early recognition, support for multidisciplinary care and expanded access to emerging therapies. Only by addressing these unmet needs can we begin to improve the outlook for patients and families affected by rare kidney diseases.

The NHS and rare kidney diseases

People with rare kidney diseases face unique challenges that existing NHS pathways do not fully address. There is a lack of specialised infrastructure needed to provide quality care to people with rare kidney diseases. The government's 10 Year Health Plan for England sets out a long-term vision for more integrated and personalised care but does not specifically address kidney disease. As a result, significant service gaps are likely to remain in meeting the specific needs of kidney patients, particularly in areas such as timely diagnosis, transition between paediatric and adult services, and access to specialist care, unless more targeted policy interventions are developed and delivered. To illustrate efforts aimed at addressing these gaps, key initiatives supporting people with kidney diseases in the UK are highlighted here: the UK Rare Diseases Framework, the renal clinical networks, national health strategies and the British Association for Paediatric Nephrology (BAPN).

UK Rare Diseases Framework

The UK Rare Diseases Framework aimed to enhance outcomes for individuals with rare diseases – including rare kidney diseases – through four key national priorities: expedited diagnosis, enhanced professional awareness, improved care coordination and increased access to specialist treatments.[32] These priorities are vital for addressing the complex, lifelong needs of people with rare kidney diseases.

This Framework has led to progress in several areas relevant to rare kidney diseases, including the expansion of genetic testing, the development of rare disease education tools for clinicians and efforts to streamline access to orphan drugs.[32]

Despite some progress, key implementation challenges remain.[32] Access to specialist clinics and centres with expertise in rare kidney diseases remains uneven across regions. Some areas lack dedicated rare disease or adolescent nephrology services, leading to variation in the quality and availability of care. Inadequate referral pathways and poor signposting also result in delayed diagnoses and missed opportunities for clinical trials or access to relevant treatments.[33–35]

These system-level gaps limit the ability of the NHS to fully realise the objectives of the Framework for people living with rare kidney diseases.[32] With the current UK Rare Diseases Framework set to expire in 2026, it is vital the Government develops a new strategy to address this important policy area.

Renal clinical networks

In England and Wales, the renal clinical networks are an initiative designed to improve the delivery and coordination of kidney care.[36] Their core mission – bringing together primary care, hospitals, specialists and community services – is to build a connected system that enables timely, equitable and high-quality care for individuals with CKD and acute kidney injury (AKI), regardless of their geographic location.[36]

By integrating services across the care pathway, the renal clinical networks aim to:

- Reduce delays in diagnosis and treatment
- Improve continuity of care as patients move between providers
- Promote equitable access, regardless of where a patient lives, and address health inequalities
- Enhance collaboration between healthcare professionals

Although not explicitly designed for rare kidney diseases, their coordinated approach offers a foundation for improving care in the area. People with rare kidney diseases often face fragmented services and long diagnostic journeys.[4] One key reason is that many of these diseases do not only affect the kidneys but also involve other organs, requiring input from multiple medical specialties. The networks' emphasis on integration and collaboration could help reduce diagnostic delays and support consistent treatment pathways for people with rare and complex needs.[36] However, there are still important limitations in the scope when it comes to coordinating care for rare diseases. The networks currently offer no dedicated provision for people with rare kidney diseases and no specific training for healthcare professionals to address their unique challenges. Furthermore, little evidence is available to assess whether these networks are achieving their aims for rare kidney diseases in practice. Without targeted evaluation and adaptation, it remains uncertain whether they are delivering meaningful improvements for this group.

Another major gap is the exclusion of children. Children with rare kidney diseases fall outside the scope of the networks, creating gaps in care and challenges during transition to adult services. This responsibility falls to the NHS England Paediatric Renal Service and the Welsh Clinical Network for Paediatric Nephrology.[36] In Scotland there is a Scottish Paediatric Renal and Urology Network but no equivalent renal network for adults. In Northern Ireland, there is no specific rare kidney network for either adults or children, and care falls under the broader Rare Diseases Action Plan led by the Northern Ireland Rare Diseases Implementation Group. More targeted investment is needed.

National health strategies

The 'Fit for the Future: 10 Year Health Plan for England' promises to transform the NHS through three shifts: hospital to community, sickness to prevention, and analogue to digital. Although the Plan makes only limited reference to kidney disease overall – and does not specifically address rare kidney diseases – it does include a welcome mention of kidney care within the genomics section. However, further policy should be developed which clearly defines care pathways to support those affected, including the need for earlier screening and detection.

In Scotland, the government is drafting a Long-Term Conditions Framework which should include ambitions for improved outcomes in kidney disease. In Wales, the Quality Statement for kidney disease sets out what good care should look like.

British Association for Paediatric Nephrology (BAPN)

The BAPN is an inclusive community of professionals dedicated to advancing care for children and young people with kidney disease. In collaboration with the UK Kidney Association, it provides strategic leadership in clinical practice and research, encouraging collective engagement to shape the future of kidney care.[37] Its aims are to improve the standard of kidney care and advance clinical expertise by promoting collaborative research and quality improvement initiatives.[37] As much of the kidney disease experienced by children and young people is rare, the BAPN plays a leadership role in sharing knowledge and best practices across the NHS in all four nations.

International perspective

A resolution formally recognising kidney disease as a major and growing public health issue was passed at the 78th World Health Assembly. It urges countries to integrate kidney care into national health strategies; expand prevention, early detection and treatment efforts, and strengthen primary healthcare services. This global commitment is especially relevant for rare kidney diseases, which often go undetected or are mismanaged due to lack of infrastructure and clinical awareness.[38,39]

A further resolution on rare diseases was also adopted recognising rare diseases as a public health priority and calls on the World Health Organization to develop a 10-year Global Action Plan to improve diagnosis, access to care and social inclusion.[38,39]

Bridging the gaps

While the renal clinical networks, BAPN, UK Rare Diseases Framework and other recent initiatives have laid important groundwork, they have not yet fully addressed the specific needs of people with rare kidney diseases. Major and unacceptable gaps remain – not just in service provision, but in how these initiatives are implemented and aligned. For example, rare disease pathways are often not embedded in mainstream renal care, and few mechanisms exist to ensure consistent delivery across regions. The lack of integration between paediatric and adult services continues to leave young people particularly vulnerable during transition. Moreover, while national frameworks emphasise early diagnosis and equitable care, people with rare kidney diseases still frequently face diagnostic delays and limited access to specialist centres. Addressing these shortcomings requires more than broad ambition: it demands sustained investment in specialist services, stronger links between initiatives and targeted policies that explicitly include and monitor rare kidney diseases. Ensuring equitable, high-quality care for this underserved population must be a priority in future NHS planning and resource allocation.

> "I'd got talking to a mum who has a little boy with a similar story to me, just 20 years apart. He's about ten now but also was diagnosed with IgA vasculitis and within five months his kidneys had failed – the same as me in 1999. It became clear that the treatments and outcomes hadn't really changed in 20 years, and I was shocked. Hopefully in the next 20 years or less, what I've experienced since childhood will be a thing of the past, because I don't want anyone to go through what I have."
>
> **Kathryn Croker, diagnosed with IgA vasculitis at 13 years old**

Research and innovation in rare kidney diseases

The treatment landscape for rare diseases is evolving, driven by advances in diagnostics, therapeutics and clinical practice. While treatment approaches have primarily focused on managing symptoms, delaying disease progression and preventing complications, newer approaches are targeting the underlying causes of individual diseases. Despite growing treatment options for some rare diseases, targeted treatment options for most rare kidney diseases are still very limited. Other challenges that still exist are delays in recognition and diagnosis, high costs and difficulty in accessing care.

Rare kidney diseases treatment landscape

Treatment options for the majority of rare kidney diseases are limited to supportive and symptom management using blood pressure control medications such as inhibitors of angiotensin converting enzyme, or ACE, angiotensin receptor blockers, or ARBs, and broad immunosuppressants (where appropriate), focused on slowing disease progression and preventing complications.[23, 24, 40] While these medications have been essential to managing people with rare kidney diseases for decades, they are non-specific, do not address the underlying causes of the disease and can cause side effects.[4]

Notably, for a select few rare kidney diseases, the introduction of disease-specific therapies has improved patient outcomes. For instance, cystinosis has been transformed from a fatal childhood disease to a chronic condition thanks to the introduction of the cystine-depleting therapy, cysteamine, in the 1990s. While cysteamine was a breakthrough, it is a lifelong therapy that requires strict, frequent dosing, which imposes adherence challenges on affected individuals and their carers. The medication also causes body odour, bad breath and nausea, which impact quality of life and potentially further hinder adherence.

Crucially, cysteamine does not cure the underlying genetic defect, meaning many people with cystinosis still progress to kidney failure and need dialysis and/or a kidney transplant, experience multi-organ complications, and have a shortened life expectancy of 40–50 years.[25, 41–44] While cysteamine is a crucial, albeit difficult, disease-specific treatment, it puts people with cystinosis in a better position than those with other rare kidney diseases. For them, there are limited options beyond symptom management before their kidneys fail. There is a monumental task ahead to develop desperately needed disease-specific, effective treatments across the full spectrum of rare kidney diseases.

Targeted therapy is a type of treatment that specifically addresses the underlying biological mechanisms – such as genes, proteins or pathways – that contribute to the development or progression of a disease. Unlike generalised treatments, which can affect many cells throughout the body, targeted therapies aim to act only on the disease-causing cells or processes, reducing damage to healthy tissues and ideally resulting in fewer side effects and improved effectiveness. A shift in focus towards developing targeted therapies is defining clinical innovation in many disease areas, including for some rare kidney diseases.

Developing targeted treatments for rare kidney diseases poses a unique combination of scientific, clinical and logistical challenges that go beyond those seen in more common kidney conditions. The kidney's complex structure – comprised of over 18 specialised cell types – and its highly selective filtration system make it difficult to deliver therapies precisely to affected areas, especially using large molecules or gene-based treatments.[23] Many of these diseases have poorly understood or highly variable biological mechanisms and vary widely in their clinical presentation, complicating the development of targeted therapies. Moreover, these conditions often begin in childhood, requiring age-appropriate dosing and formulations and long-term safety data, making it difficult to measure meaningful changes over the short timelines typical of clinical trials. Small and geographically dispersed patient populations limit trial recruitment and statistical power. Furthermore, there are few validated biomarkers or surrogate endpoints to track disease progression or therapeutic response, adding uncertainty to trial outcomes.

Pathways for the development of new treatments

Developing new treatments for rare kidney diseases relies on a complex ecosystem and multiple stakeholders, including scientific and clinical researchers, pharmaceutical companies, regulatory agencies and policymakers.[45,46] Randomised clinical trials (RCTs) are the gold standard and rely on having at least two arms to compare against each other (i.e., one group of patients receiving the new medicine and the other receiving standard treatments or placebo) and large numbers of patients to show statistically significant benefits. These are largely suited to adult patient populations with more common diseases, so conducting research in rare and paediatric populations presents distinct challenges.[27] In settings where there are no available treatments, for example, it may be considered unethical to randomise participants to a control group.

Rare kidney disease research therefore requires novel methodology that is designed to meet its unique needs. As an example, common measures in kidney trials like estimated glomerular filtration rate (eGFR; the rate at which blood is filtered by the kidneys) or proteinuria (amount of excess protein found in the urine) may not change significantly during the trial period.

The adoption of modern adaptive trial designs must be considered, with the support of the regulatory authorities. These should also include new, more appropriate surrogate endpoints, which may be harder to validate using traditional statistical methodology, and the adoption of new biomarkers where possible.

Furthermore, overcoming the challenges associated with conducting research in children, how to incorporate small and clinically diverse patient populations, and getting therapies through to patients despite their low market value to drive change are all vital considerations.[27, 45, 46] International recruitment is often needed to reach sufficient numbers, which poses additional logistical challenges.

The paediatric onset of many rare kidney diseases introduces additional complexities.[25, 27, 45, 46] Young children are often unable to report symptoms themselves, necessitating the use of clinician- or observer-reported outcomes, which may lack the sensitivity and standardisation needed for regulatory acceptance. Diagnostic delays, limited understanding of disease natural history and a lack of validated surrogate endpoints further hinder trial design and execution.[45, 46] Moreover, for paediatric kidney diseases, early treatment is especially crucial because there is often a limited window of time during which starting medication has the greatest impact on long-term outcomes.

> Although receiving an earlier diagnosis would not have changed the outcome, it would have allowed me earlier access to medical interventions that would have made life more enjoyable. I would like to see a world that offers children the same access to prevention, protection and treatment as adult kidney patients have. You have the power to hand us back our childhoods."
>
> **Charlie Frieland, 15-year-old with nephronophthisis**

In the UK, once a new medicine enters the clinical development phase, the Medicines and Healthcare products Regulatory Agency (MHRA) has the responsibility of reviewing clinical evidence and determining whether it meets the required standards for safety, efficacy and quality.[47] For orphan medicines, defined as those targeting conditions affecting fewer than 1 in 2,000 people, the MHRA also considers whether the treatment addresses an unmet need or provides a significant benefit over existing options. Once a treatment is approved, pharmaceutical companies may receive certain regulatory incentives to encourage the development of medicines for rare or underserved conditions. These incentives can include a period of market exclusivity (when no other company can sell the same medicine) or extensions to existing patents (e.g., if the company has completed paediatric investigation plans). These incentives are intended to promote innovation, but they can also delay the entry of lower-cost alternatives, which may impact patient access over time. Table 1 illustrates some of the barriers or challenges in rare disease drug development.

Table 1. Challenges in rare disease drug development

Discovery and investment	Small patient population	Limited commercial incentives	High R&D risk
Clinical trials	Lack of natural history data	Ethical issues with placebo control	Few eligible patients
Regulatory approval	Need for surrogate endpoints	Limited data for review	Multiple, unaligned regulatory systems
Reimbursement	High upfront treatment cost	Payer concerns about value	High impact on annual budgets, especially for single treatments
Post-launch and uptake	Systems readiness	Need for specialist delivery models (e.g. for gene therapy)	Slow adoption

Following regulatory approval, bodies such as the National Institute for Health and Care Excellence (NICE) in England and the Scottish Medicines Consortium (SMC) assess whether the NHS should fund a treatment.[48] Their evaluations focus on value for money (termed cost-effectiveness), which compares the extra cost of a new treatment with its benefits. This traditional assessment framework is primarily built around evidence from large RCTs and is therefore not well-suited to rare conditions.

To address this, some specialised assessment routes have been introduced, such as NICE's highly specialised technology (HST) programme and Scotland's ultra-orphan pathway. These appraisal routes are designed to accommodate the evidence limitations and uncertainties inherent in rare disease research.[49,50] However, HSTs have restrictive inclusion criteria and there is limited annual capacity for these appraisals, which means that many rare disease therapies still undergo standard evaluation processes that may not fully accommodate the complexities of rare disease research and are therefore less likely to receive a positive recommendation.[51,52] One study examining whether NICE demonstrated flexibility in evaluating treatments for rare diseases compared six recent single technology appraisals (STAs) for rare diseases with six for common diseases.[52] Rare disease technologies were generally subject to longer appraisal times (an average of 504 days vs 386 days for common STAs), spent 48 days longer in NICE, and required more appraisal committee meetings (2.0 for rare vs 1.3 for common).[52] It is difficult for drug manufacturers to prove that a rare disease treatment offers good value for money within the strict £20,000–30,000 cost-effectiveness threshold. This extended timeframe potentially leads to delays in people living with rare diseases accessing life-changing treatments.[52] Addressing these barriers remains critical to ensuring timely access to innovative treatments.

Clinical pipeline and noteworthy trials

Research into rare kidney diseases is particularly pressing due to the high unmet need and profound impact these conditions have on individual people, many of whom are children, their families, and the wider healthcare system.[5,24] Investment in innovation and research has the potential to uncover novel biological mechanisms that could inform new treatments for both rare and common kidney diseases, ultimately alleviating some of the burden associated with these conditions. Advances in rare kidney disease research also drive the development of personalised medicine, enabling more precise and effective therapies tailored to each person's unique genetic and molecular profile.

The global clinical pipeline for rare kidney diseases includes dozens of potential new therapies, including monoclonal antibodies, small molecules and gene therapies, that target various biological pathways. Currently, the majority of clinical trials for rare kidney diseases focus on IgAN, lupus nephritis (LN) and ADPKD, many of which target the underlying dysregulated pathways that cause the diseases.[24] For IgAN and LN, most target the dysregulated immune pathways (abnormal immune system responses) that contribute to kidney damage.[24,53]

The evolving treatment landscape for IgAN has been particularly notable, with the first disease-specific therapy, a formulation of budesonide (a corticosteroid), approved in the US in 2021 and the EU in 2023. It works mainly in the gut to reduce the activity of overactive immune cells that make harmful Gd-IgA1.[54,55] Sparsentan, a dual-action endothelin and angiotensin II receptor antagonist (a drug which prevents blood vessel constriction and reduces pressure in the kidneys), was approved for IgAN in adults in the US in 2023 and recommended by NICE in May 2025.[56] Sparsentan is also being evaluated in adults and children with focal segmental glomerulosclerosis (FSGS) and other glomerular diseases, but initial results are disappointing.[56] While sparsentan was effective at reducing proteinuria, there was no significant difference between it and irbesartan, a blood pressure–lowering medicine, in slowing the decline of kidney function.[57] Iptacopan, which targets a part of the immune system called the complement system, was also approved for IgAN in adults in the US and the EU in 2024, but is not yet approved for children.[58]

Research and treatment for children with rare diseases consistently lag by about 10–13 years behind those for adults, even for therapies with demonstrated long-term efficacy in adults.[25] This delay is illustrated by the case for IgAN: in July 2025, 196 interventional studies were registered on ClinicalTrials.gov, yet only 33 of these included children.[59,60] Consequently, many therapies are not licensed for children or are used off-label, preventing the collection of rigorous evidence for efficacy, safety and dosing. As children are the population with the greatest potential for long-term benefit, there will be a generation of people with CKD for whom earlier access to medication could have changed their life trajectory.

Gene therapy is emerging as a promising approach for treating adults and children with rare kidney diseases, many of which are monogenic (caused by single gene mutations).[23] These therapies aim to correct the underlying genetic defect, offering the potential for long-term or even curative treatment. Farabursen is a novel next-generation oligonucleotide (short chain of DNA) being evaluated for the treatment of ADPKD.[61] Preliminary data suggest that targeting pathways involved in cyst growth, which contributes to the decline in kidney function, may help slow disease progression. Nephropathic cystinosis, a rare kidney disease where early intervention is critical for children to live beyond ten years of age, may see a changed course in the lives of affected children with the advent of DFT383 (previously AVR-RD-04). This autologous hematopoietic stem cell gene therapy is currently in Phase I/II clinical trials in children aged two to five years old.[62] The gene therapy aims to be curative by genetically modifying patients' stem cells to express a 'normal' version of their defective CTNS gene, thereby offering a crucial early childhood intervention.[23]

Chimeric antigen receptor-T (CAR-T) cell therapy also represents a transformative shift for the treatment of LN, moving beyond chronic immunosuppression with non-specific medicines toward a potential one-time, intervention.[63] CAR-T cell therapy for LN involves reprogramming a patient's own immune cells to target and eliminate the overactive cells that are attacking the kidneys, to reduce inflammation and prevent further damage.[63] Multiple early-phase trials are underway with promising preliminary data.[64,65] However, key challenges remain, including costs (the list price of CAR-T before NHS discount is over £280,000 per infusion),[66] manufacturing scalability and uncertainty surrounding long-term outcomes.

The treatment landscape for rare kidney diseases needs to rapidly evolve to avert the kidney failure crisis, driven by scientific advances and a growing recognition of the urgent need to improve outcomes. Novel therapies, such as targeted small molecules, immune pathway modulators, gene therapies and CAR-T cell approaches, are shifting the focus from symptom management toward more targeted and potentially curative treatments. Despite this progress, the majority of rare kidney diseases still lack effective or approved treatments, and children in particular continue to face considerable delays in access to innovative solutions. To truly transform care, a dedicated focus on understanding underlying disease mechanisms is needed to use biology to guide better diagnostics, treatment and tailored management. This may include high-throughput discovery science obtained by whole-genome screening and supported by national rare disease biobanking, or the use of artificial intelligence to screen diagnostic imaging or perform virtual drug screening to personalise treatments.

Figure 2. Innovation in the rare kidney disease treatment landscape is characterised by a shift to targeted therapies.

Targeted therapies			
Improved versions of existing therapies	Immune pathway modulators	Gene- and RNA-based therapies	CAR-T therapies
Targeted-release budesonide (a corticosteroid) approved for IgAN Sparsentan approved for IgAN, ongoing phase 2 trials for FSGS, IgAV, and Alport syndrome Imlifidase for anti-GMB (phase 3)	Iptacopan approved for C3G and IgAN (UK approvals pending) Atacicept for IgAN (phase 3) Ravulizumab for CSA-AKI, IgAN and aHUS (phase 3) Inaxaplin for AMKD (phase 3)	Rarabursen microRNA-17 inhibitor for ADPKD (phase 1b) Lademirsen, an anti-microRNA-21 therapy, for Alport syndrome (phase 2) DFT383 HSC therapy for cystinosis (phase 1/2) ABO-101 for hyperoxaluria (phase 2/3) Nedosiran for hyperoxaluria (phase 2)	KYV-101 for LN (phase 1/2) Rapcabtagene autoleucel for LN (phase 2)

Accelerating innovation in the UK

As the potential clinical pipeline expands, continued research and innovation, which are key to delivering better treatment options to people with rare kidney diseases, must also expand.

The UK is well positioned to accelerate clinical trial setup by leveraging the continuity and integration of the NHS, potentially enabling faster and more coordinated trial delivery than in other systems, including those in Europe and the US. Embedding clinical research within NHS services offers clear benefits: it enables earlier access to innovative therapies for patients with limited options and generates evidence to support clinical practice and regulatory decisions.[6,67] Over time, this could potentially reduce the impact on NHS services by helping to delay or avoid costly interventions such as dialysis and kidney transplant.[6,68]

The UK's regulatory environment is in a position to evaluate new therapies based on global and local evidence.[69] In practice, however, delays persist in the timely approval and access of rare disease treatments.[52,69] NICE's HST pathway, although designed for rare diseases, remains narrow in scope, meaning many rare diseases are evaluated in the STA pathway, where they are less likely to receive a positive recommendation.[52,69]

Meaningful patient involvement is crucial in rare kidney disease research and access decisions, especially when clinical evidence is limited. However, HTA processes, particularly at NICE, place a disproportionate burden on small patient advocacy groups to provide input. Many groups dedicate considerable resources and energy to supporting services and navigating these processes, often without support or recognition. These hidden costs are typically borne by small charities, volunteers and unpaid advocates who frequently self-fund their participation and face emotional pressure from their communities. Without dedicated support and independent funding for meaningful engagement, the patient voice risks becoming limited to well-resourced diseases.[70]

Importantly, there are unacceptable variations in access to treatment and research across the UK. A clear 'postcode lottery' exists, with some individuals benefiting from specialist services and clinical trials while others are left behind due to geographic or service-level inconsistencies. This disparity extends to transitions between paediatric and adult services, where poorly coordinated transitions to adult care risk undermining continuity of care and patient engagement.[6]

Innovation in the management of rare kidney diseases extends beyond medicines. Advances in genomic sequencing are enabling earlier and more accurate identification of genetic and molecular causes of disease and are supporting the development of personalised treatment plans.[6,23,24] Digital health tools (including the NHS app, telemedicine and remote monitoring) offer new ways to connect people with rare kidney diseases with specialist care and clinical research, particularly for those living far away from major centres.[71–73]

The UK's research ecosystem is well placed to lead the way in rare kidney disease research. Initiatives such as the LifeArc-Kidney Research UK Centre for Rare Kidney Diseases, RaDaR and the National Unified Renal Translational Research Enterprise (NURTuRE) are fostering collaboration between academia, industry and clinical experts.[9,68,74,75] These efforts are largely charity funded, but sustained government investment and national coordination are needed to fully realise their potential.

Spotlight on four rare kidney diseases

Rare kidney diseases encompass approximately 150 conditions affecting about 160,000 people in the UK,[6,10,76] 80 with distinct causes, patient populations (with 34 linked to specific ages or ethnic backgrounds), symptoms and progression. To illustrate the heterogeneity of these diseases, four examples are highlighted, providing a different view of the world of rare kidney diseases:

Autosomal dominant polycystic kidney disease (ADPKD)

Cystinosis

Congenital anomalies of the kidney and urinary tract (CAKUT)

IgA nephropathy (IgAN) and IgA vasculitis (IgAV)

Each overview has a firsthand account from people or carers of people living with these diseases, followed by a clinical perspective and brief examination of the disease, including available data on the economic, human and societal impact associated with it.

Autosomal dominant polycystic kidney disease (ADPKD)

ADPKD is the most common inherited kidney disease, typically manifesting in adulthood (ages 30–40) and caused by mutations in the PKD1 or PKD2 genes.[77] In the UK, ADPKD affects approximately 1 in every 2,000 people, which equates to around 34,000 people.[10] People with ADPKD develop many fluid-filled cysts in their kidneys that can make them enlarge and impair their function, which can lead to high blood pressure, pain, blood in the urine and eventually kidney failure.[78] Other reported symptoms include fatigue, shortness of breath, weakness and bloating.[79] The disease is often invisible until later stages, and progression to kidney failure varies depending on which genetic variant a person has.[6] The median age at diagnosis is 38, and the median age of kidney failure is 59.[6]

The introduction of tolvaptan in 2015[80] marked the first and only disease-modifying treatment for ADPKD. It has been demonstrated to slow the growth of kidney cysts and preserve kidney function in select patients but has considerable side effects. Real-world data show that about one in three people with ADPKD stopped taking it within one year; more than half (56%) stopped taking it by three years.[81] A similar study in Italy found that 18% of 122 people treated discontinued treatment due to kidney failure (6%), frequent urination (3%) and family planning (3%).[82] Among 220 people studied in Spain, 78% completed one year of tolvaptan. Reasons for discontinuing treatment included frequent urination (11%), worsening kidney function (5%) and liver toxicity (2%).[83] While tolvaptan can delay reaching kidney failure by one year for every four years on medication, this benefit is modest compared to the side effects.[80,84,85]

The costs associated with ADPKD are substantial due to the need for ongoing care and, in later stages, dialysis or kidney transplant. Dialysis costs the NHS about £1.05 billion annually.[10] As ADPKD accounts for approximately 10% of patients requiring kidney replacement therapy,[86,87] an estimated £105 million of these costs are attributable to ADPKD.[99] an estimated £105 million of these costs are attributable to ADPKD.[10,86,87] Dialysis for all rare kidney diseases (including ADPKD) costs the NHS approximately £263 million a year.[10]
For those taking tolvaptan, the annual fee estimated by the manufacturer is £15,750 per person, before NHS discounts, which is cheaper than dialysis.[80] ADPKD also imposes an indirect economic burden through loss of individual productivity: people with ADPKD have a 64% chance of being out of work by the time they need kidney replacement therapy.[88] Because symptoms are often invisible, some people with ADPKD worry that their employers may not recognise it as a disability, which could lead to them being laid off, financially insecure and unable to access disability support.[79,89] John, whose story appears later, had to stop working 14 years before he reached retirement age. Family caregivers for people with ADPKD are also affected; one in four (27%) experienced lost working hours or employment disruption due to their caregiving responsibilities.[90]

Added to this are the psychological and social impacts on people living with ADPKD and their carers. Receiving a diagnosis of an incurable disease that could be passed on to children has been described by some as a "bomb."[79] Some described feeling helpless or hopeless – frustrated by the limited information, uncertain about when kidney replacement therapy might be needed or whether to have children.[79] In a survey of 349 people with ADPKD in the UK, 22% screened positive for clinical depression and 62% reported feeling guilty about passing the disease on to their children. Those with more advanced disease reported poorer quality of life and increased psychosocial burden.[91]

A qualitative study of 33 teenagers with ADPKD across 13 countries found that 18% of participants missed school at some point, which could potentially limit their career prospects in the future.[90,92] Some reported feeling uncomfortable or self-conscious because of the need to urinate frequently.[93]

John and his family's journey

John Roberts, 67, a clinical supervisor from Manchester, had to give up work due to his ADPKD. He and his son were both diagnosed in 1995 after his son experienced severe migraines. "I remember being totally confused… What I read frightened me, as I'd already passed the life expectancy for someone with the condition."

By 2014, John's kidney function had deteriorated to the point that he needed dialysis. The pain from the growing cysts in his kidneys forced him to stop working, creating major financial strain. In 2017, he developed another rare and painful condition called calciphylaxis, which he survived despite initial recommendations for leg amputation. His son, however, has so far been affected very little by his disease and has been taking tolvaptan.

In 2018, after four years on dialysis, John's daughter surprised him by offering to donate her kidney. "After my daughter left, I just burst into tears thinking about the gift she was giving me and how it would transform my life."

Though John has experienced complications, such as post-transplant diabetes, his new kidney has given him the chance to enjoy life again and spend time with his grandchildren and great-grandchildren. "Maybe one day there will be a tablet we can take to cure kidney disease."

Gareth and Caroline's story

Caroline Prodger's life changed forever in March 2025 when her husband, Gareth, died from multiple organ failure following an infection. He had been on immunosuppressive drugs after receiving a heart transplant, which became necessary due to a rare complication of his rare kidney disease, ADPKD.

They had met 30 years earlier at university. "He was super bright, funny and kind," she recalled. But even from the beginning, their relationship was shaped by Gareth's inherited kidney disease. His father had lived with ADPKD for years and spent over a decade on dialysis after two failed kidney transplants.

Gareth was diagnosed shortly after meeting Caroline. They focused on controlling his blood pressure, a strategy known to slow the progression of ADPKD. They held out hope for new therapies, including the drug tolvaptan, which Gareth began taking later in life. Tolvaptan is not a cure for ADPKD; on average, for every four years taken, it delays kidney failure by one year, but it requires significant adjustments to daily life as people taking it need to urinate often and struggle to drink enough water.

But in 2020, Gareth's health suddenly declined. He began experiencing breathlessness and would often wake up struggling to breathe. He was later diagnosed with a serious heart condition, dilated cardiomyopathy.[94] Plans were made for a heart transplant, followed by a kidney transplant from his brother. During that time, he suffered several setbacks. First, he had to begin peritoneal dialysis at home. Then he needed hernia surgery and suffered a mini-stroke. In October 2023, he developed peritonitis and had to have haemodialysis four times a week at Royal Berkshire Hospital, where the care team worked diligently to keep him well enough for a transplant. Though the heart transplant was successful in 2024, his body was too weakened to undergo kidney transplantation. He died in March 2025, aged 50, after developing sepsis.

Clinical perspective

Dr Matt Gittus, Specialist Nephrology Fellow

John's experience demonstrates the unpredictability of ADPKD; disease progression can vary widely, even between individuals in the same family carrying the same genetic variant.[79, 94–96] His son was diagnosed during childhood, while John remained symptom free as his disease quietly progressed for many years. His son is doing well on tolvaptan, whilst John has had dialysis, received a kidney transplant and now lives with diabetes. This variability makes it challenging for clinicians to predict disease progression or to provide the information individuals need to make informed decisions about family planning. Additionally, John was unlucky as calciphylaxis is not directly related to ADPKD, but dialysis is a risk factor.[97] Perhaps if a more effective treatment had been available, it would have delayed the need for dialysis. A better treatment may have also enabled him to keep working. Pain is the main reason people with ADPKD cannot work or do things that they enjoy.

Advances in imaging and genetic testing have improved early diagnosis of ADPKD, but their use in clinical practice remains inconsistent. Moreover, interpreting genetic variants remains a challenge, and roughly 6% of patients with ADPKD have no identifiable genetic cause.[94, 95, 98, 99] Beyond clinical challenges, the psychosocial toll is profound. Many patients live with anxiety about their future and whether their children will inherit the disease. The lifelong burden of care, including dialysis, kidney transplant and ongoing monitoring, places substantial strain on healthcare resources and results in high long-term costs.

Cystinosis

Cystinosis is a rare, life-threatening metabolic disorder that is typically diagnosed in early childhood by around age two.[44,100,101] In the UK, cystinosis affects between 1 in 200,000 and 1 in 100,000 people, which equates to approximately 340 to 680 individuals nationwide. Only two to three new cases are diagnosed each year.[102] There are three types of cystinosis, with nephropathic cystinosis (NC) being the most common and severe form.[43,44,103] 95% of people living with cystinosis have NC, which usually starts in early childhood. The other two are intermediate cystinosis, beginning in adolescence, and non-nephropathic ocular cystinosis, which only affects the eyes and typically begins in adulthood.[44,103] NC (for simplicity, referred to as cystinosis throughout this report) is caused by a defect in the CTNS gene that prevents the body from properly removing a substance called cystine.[44,100,101] Cystine is a natural waste product, but in people with cystinosis, it accumulates inside tiny storage compartments in cells called lysosomes.[104] Over time, this buildup primarily damages the kidneys and eyes, but it can also affect the muscles, pancreas, thyroid and white blood cells.[104] Children with cystinosis may be smaller than their peers and have soft or weak bones.[44] Symptoms typically begin around six months with frequent urination, constant thirst and dehydration.[104] Children with cystinosis go through puberty one to two years later than other children their age, and males are infertile. Most children with cystinosis reach kidney failure by age 15.[101,105]

The management of cystinosis involves lifelong, intensive use of health services.[41,42] Regular blood tests are necessary to monitor cystine levels and adjust medication dose accordingly.[106] These tests must be conducted at one of five certified labs located at the following hospitals: Birmingham, Great Ormond Street, Leeds, Manchester Hospital and Evelina London.[102,107] In addition to cysteamine, people with cystinosis often require additional treatments to manage other symptoms of the disease, including thyroid medication, proton pump inhibitors to alleviate stomach upset caused by taking cysteamine tablets, and various supplements to replace essential electrolytes or vitamins.[102,106]

People with cystinosis need multiple services from a team of different NHS specialists, such as kidney services (including kidney replacement therapy); eye care to prevent impaired vision; neurology service to monitor for brain damage and breathing problems; speech and language services for those with difficulty swallowing; nutrition, dietetics and endocrinology for supplementation and diabetes management as needed; bone services for calcium and phosphate lost in urine; sexual and fertility treatment; genetics and laboratory services for regular lab tests, diagnosis and genetic counselling; and psychology services, as cystinosis can markedly affect schooling, the ability to get a job and social lives.[105,106] Patient support groups may also help families navigate treatment, share practical tips and advocate for more research.

Children with cystinosis often experience both physical and emotional challenges. Common issues include tiredness, anxiety, depression, attention difficulties and aggressive behaviour. They may be bullied for the medication effects or their short stature. Having to take medication multiple times a day can make it difficult to hang out with friends. While they generally have normal IQs, some studies suggest there may be subtle academic differences. For example, two studies indicate children with cystinosis have poorer performance in arithmetic and a trend toward poorer performance in spelling. This effect may be reduced with early cysteamine treatment.[108, 109]

Morven and Alex's story

Fifteen-year-old Morven lives with cystinosis. Diagnosed in infancy, they now follow a strict, round-the-clock medication schedule to slow the progression of kidney disease. Their medication, mercaptamine, is taken orally every six hours, including during the night, and disrupts sleep and daily life. While the treatment helps protect their kidneys, it comes with side effects like nausea and a strong, egg-like smell that can cause bad breath and body odour. This can be embarrassing and may lead to teasing or social exclusion. At school, Morven must visit the first aid room during breaktimes to take their medication, missing time with friends and sometimes feeling isolated as a result.

Morven's mother, Alex, says the burden of care is constant. Mercaptamine must be taken on time and under specific conditions. Morven also takes mercaptamine eyedrops, which must be refrigerated, complicating travel and school life. Despite these challenges, Alex is grateful that Morven does not yet need dialysis or a kidney transplant, which doctors initially predicted would happen in early childhood. "It's hard to know what to expect with a rare disease, because every case is so different and there are too few to compare."

The family's journey with cystinosis began unexpectedly. Morven was diagnosed at eight months old during unrelated blood tests. Alex recalls the overwhelming wave of medical information that followed. While Morven has avoided some complications, such as a gastrostomy tube and extended hospital stays, the disease still impacts nearly every part of their daily lives.

"Morven's is one story amongst many, all of which are individual. There are stories which are even more heartbreaking, and I want to acknowledge that. It's important for researchers, clinicians and families affected by cystinosis to work closely together to find breakthroughs which can transform lives", Alex says.

Julia and Kirsty's story

Scottish seven-year-old Julia also lives with cystinosis. Diagnosed at age two after months of distressing symptoms and repeated visits to the GP, her family's story highlights the difficulty of getting a timely diagnosis and the damage delays can cause. Julia's mum, Kirsty, explains how treatment dominates her daughter's daily life:

"Whatever she's doing – school, friends' parties, dance class – everything gets cut short. When the medicine is due, she has to have it."

Julia takes 27 doses a day, including oral medication and eye drops taken every two waking hours to prevent the painful buildup of cystine crystals in her eyes. Treatment side effects include stomach ulcers, and Julia also struggles with leg pain, tiredness and frequent urination, which can be embarrassing at school.

She loves dancing, singing and designing clothes for her dolls, but she's starting to realise her life is different from that of her friends. "She has cried a lot, questioning why this is happening to her," Kirsty says. "She's a typical wee girl... [but] she's starting to feel self-conscious."

The road to diagnosis was long. Julia had stopped growing, lost weight and developed grey, sunken eyes. Her mum was advised to restrict fluids, something later found to have harmed her kidneys. "It was the most horrific time," Kirsty recalls, "She would shake a lot and vomit regularly. I was begging the GP to send us to a specialist urgently." All this was happening while Kirsty was also caring for her terminally ill father.

Eventually, a gastrostomy tube (a small tube used to provide liquid feed) was inserted to help Julia gain weight and ensure medication delivery. It took nearly a year for Julia to adjust to her medication. She was frequently hospitalised and missed so much nursery that she had to repeat a year of school.

Now, with Kirsty working onsite at the school, she's able to give Julia her medication during the day, something that's helped reduce how often Julia needs to be admitted to hospital. At home, her parents spend hours preparing feeds and cleaning equipment. The routine is exhausting, and treatment remains difficult for Julia, who is also in therapy to help manage the medical trauma she's experienced.

Julia is likely to need a kidney transplant within the next five years, and several family members have offered to donate. "We all want to avoid dialysis," Kirsty says. The family is hopeful that new research may offer better options. They emphasise the need for increased awareness and funding to enhance care and outcomes for children like Julia.

Clinical perspective

Dr Ben Reynolds, Consultant Paediatric Nephrologist

Cystinosis is a truly devastating disease with a very high medication burden. Many families reorganise their lives to manage the demanding medication schedule, and unfortunately it must be done to preserve kidney function as long as possible. Most children require a nasogastric tube and subsequently a gastrostomy to help maintain this strict dosing schedule overnight.

For children and teens, the medication effects can be a source of embarrassment or bullying. Over time, this can have a profound impact on self-esteem and mental health. As patients grow up, the medication can affect romantic relationships, as the taste is obvious when kissing. Males with cystinosis are infertile, which brings its own emotional impact in adult life. [43, 110]

The eye drops add another layer of complexity. Cystine crystals that accumulate in the eyes make them highly sensitive to light. [43, 110, 111] The eye drops can help to dissolve the crystals, but they must be taken every day, several times a day, for the rest of the person's life. With very young children like Julia, this can be difficult and may require the assistance of two adults to administer, especially if using the licensed version, which has a thick consistency and stings on contact. [43]

Even with the best care, Morven and Julia will likely develop kidney failure. [43, 112] Cystinosis causes a 'leaky kidney' syndrome where the kidneys lose the ability to retain salts and fluids, leading to constant thirst and a need for salt replacement. [43, 112] Dialysis or a kidney transplant is often required in late adolescence or early adulthood, a critical time in education. [43, 110–112] Other complications of cystinosis include thyroid dysfunction, diabetes, bone and muscle weakness, and learning and memory problems. [110]

Morven's kidney function has lasted longer than predicted, but sadly, cystinosis is still a life-limiting condition. [43] Many patients do not live beyond their forties or fifties. Cysteamine has improved life expectancy, but research, including promising gene therapy trials, [62] is urgently needed to provide better options for everyone living with the disease.

Congenital anomalies of the kidney and urinary tract (CAKUT)

CAKUT refers to a group of birth defects that affect the kidneys and urinary system.[113] These include problems like missing or underdeveloped kidneys, blockages, or abnormal connections between the kidneys, bladder and ureters.[113] These conditions develop before birth and are the leading cause of CKD in children.[113,114]

CAKUT develops when something goes wrong during early kidney development in the womb. This can be caused by changes in specific genes or by signals between growing organs not working correctly.[115] Some children with CAKUT need surgery early in life, while others need lifelong care to monitor or treat kidney problems. In the most serious cases, CAKUT leads to kidney failure, which may require kidney replacement therapy.[116]

In the UK, CAKUT affects between 4 and 30 of every 10,000 babies born.[117] With about 695,000 babies born each year, this means that around 280 to 2,100 babies are born with some form of CAKUT.[117,118] Many of these children need ongoing care, including prenatal obstetrics, paediatric primary care, nephrology and urology. About half of all children who need dialysis or a kidney transplant have CAKUT.[119] If somehow CAKUT goes undetected in childhood, it can present with kidney problems in adulthood, as in Adam's case.

CAKUT can result in substantial costs for the NHS due to the need for surgery, ongoing check-ups and specialist kidney care. Some people living with CAKUT may self-catheterise; it is estimated that about 50,000 individuals in the UK use catheters, which is about 5 million catheters per year.[120] These statistics do not specify how many of these users are people with CAKUT, which reflects a broader lack of data on health services used by people with rare kidney diseases.

The impact of CAKUT is profound: Adam has had two kidney transplants after being on dialysis for six years, and Asher is only two years old but has already had 11 surgeries and may need multiple kidney transplants in his lifetime.

Asher's story

Two-year-old Asher Pyper was born with posterior urethral valves (PUV), a severe inherited CAKUT condition that blocked urine flow and severely damaged his kidneys in utero. Diagnosed with kidney failure shortly after birth, Asher has since had 11 surgeries, including removal of a deformed kidney. He now lives with a stoma, relies on a feeding tube and is on dialysis while waiting for a kidney transplant.

Asher's daily care is relentless. He takes more than 20 medications daily, often vomits stomach acid, and cries in pain, unable to fully communicate his discomfort. His development lags behind that of his healthy twin sister in all aspects: socially, physically and emotionally. His condition demands high-resource care: extended hospital stays, complex surgical interventions and multi-specialist teams spanning two major centres, including one over two hours from home. One parent is always by his side. That's nearly 1,250 hours spent accessing treatment since birth, half of which is just travelling to and from hospital.

The impact on the family has been tremendous. Asher's mum, Eloise, has stepped back from day-to-day management of her salon. With three other children at home, the unpredictability of Asher's needs, and frequent hospital visits, juggling childcare and family responsibilities has been difficult. Asher's sisters are distressed by the constant separation from their parents and brother, and Eloise and her husband, Ian, rarely see each other. Friends haven't met their youngest child, Effie, because she's spent most of her life in the hospital with Eloise and Asher. Ian has begun donor evaluation for a potential transplant, but the family knows it will not mark the end of Asher's treatment, only the beginning of a new phase.

Adam's story (congenital renal dysplasia)

At 21, Adam Musa was rushed to intensive care after experiencing breathing difficulties. He was diagnosed with kidney failure and needed immediate dialysis. Previously healthy and active, Adam had been showing signs months earlier: fatigue, swelling and low urine output. However, a visit to his GP did not result in further testing or the discovery of any health issues. It was only after his emergency admission that he later learned he had been born with congenital renal dysplasia, a condition where the kidneys never fully develop. The condition had gone undetected until his second kidney failed in 2009.

Adam endured months of dialysis before receiving a kidney transplant from his father. Initially successful, Adam's donor kidney failed after nine years. He went back on dialysis multiple times a week, four hours each time, for a further six years until his second kidney transplant in 2024.

Adam's care journey reflects high healthcare resource use across hospitalisations, specialist care, transplant coordination and long-term dialysis. These interventions come not only with medical strain but also economic consequences. As a husband and father of two, Adam and his family must navigate financial pressure from his limited work capacity and long-term treatment dependency. He believes an earlier diagnosis, perhaps when his symptoms appeared, could have altered his path.

Clinical perspective

Dr Lieke Hoogenboom, Consultant Paediatric Nephrologist

One of the challenges of CAKUT is its variability.[109, 121, 122] In some cases, such as Adam's, the condition isn't diagnosed until adulthood. He was born with underdeveloped kidneys, but it wasn't diagnosed until he became seriously unwell. Sadly, by the time these conditions come to light in older children or adults, there's often already been significant irreversible damage.

In other cases, such as Asher's, the effects of CAKUT are apparent from birth. His condition is severe, and he needs ongoing hospital care, surgery, dialysis and a long list of daily treatments to stay stable. Often are also bladder problems, which add further complexity. Some children can't urinate the usual way and require assistance to empty their bladders. One option, a vesicostomy, drains urine through an opening in the belly, which means they're constantly leaking and need a stoma bag or nappies. This can be tough emotionally, especially when other children their age are toilet-trained.

Another option is catheterising, either through the usual opening or a surgically created channel called a Mitrofanoff. While this doesn't leak, it still means the child manages their bladder differently, and they are constantly reminded of their medical condition.

The unpredictability doesn't just affect day-to-day care, it also means families with the same diagnosis can have vastly different experiences. One child may live a relatively normal everyday life with minimal intervention, while another with a similar diagnosis might need lifelong care and multiple transplants. This uncertainty is incredibly difficult for families, and as clinicians, we strive to support them through it with clear communication and careful planning.

We are still learning why some children's kidneys decline rapidly while others remain stable for years. Research into this area is essential because a better understanding could help us intervene earlier, reduce the burden of treatment and provide families with greater clarity about what may lie ahead.

IgA nephropathy (IgAN) and IgA vasculitis (IgAV)

IgAN and IgAV are related diseases that affect the kidneys.[123] Experts now think there are four key steps, or 'hits', that lead to IgAN. It begins when the body produces a form of IgA1 (a type of antibody) that is missing an essential sugar molecule. The immune system reacts by creating antibodies against it, which form clusters called immune complexes. These build up in a part of the kidney called the glomerulus, causing inflammation and damage which can worsen over time and prevent the kidneys from working correctly.[124]

IgAN is a common type of kidney inflammation caused by the immune system and is typically diagnosed before the age of 40.[125,126] In contrast, IgAV mainly affects children, often causes a skin rash, joint pain and stomach pain, and in some cases leads to kidney problems.[127]

In the UK, about 1 in every 50,000 people has IgAN.[128] Based on this rate, it is estimated that around 1,400 people are currently living with the condition.[125,128] IgAV affects about 20 out of every 100,000 children under the age of 17 each year.[129] Since in the UK there are around 6.5 million children aged three to ten – the age group most commonly affected – this means that there are about 1,300 new cases of IgAV in children each year.[130,131]

In addition to its direct health effects, IgAN places a measurable financial burden on individuals and families through lost productivity. Based on UK average annual earnings of £37,430 and data from the HONUS study showing an 18% productivity loss among employed people with IgAN, the productivity cost is approximately £6,650 per person per year. This includes both time off work (absenteeism) and reduced effectiveness while at work (presenteeism).[132,133] These figures underscore the broader economic impact of the disease, extending beyond the healthcare system.

Liam's story

Liam Ward was 21 when his future changed during a routine army medical examination. He had no idea he was ill until blood in his urine and high blood pressure led to a diagnosis of IgAN, a rare autoimmune kidney disease, in 2014. The diagnosis came as a shock. Liam had left university to pursue a military career and was blindsided by a chronic condition he had never heard of.

Instead of being at boot camp, Liam spent the next decade managing his blood pressure and trying to slow disease progression with medication. Over time, his kidney function declined, and he was added to the transplant list. He finally received a kidney in 2022 from a deceased donor. The transplant significantly improved his energy levels and overall quality of life. He returned to work, got married and took up running, completing the London Marathon in 2025 and raising more than £3,000 for Kidney Research UK, with the hope that individuals diagnosed with IgAN in the future will not have to go through what he did.

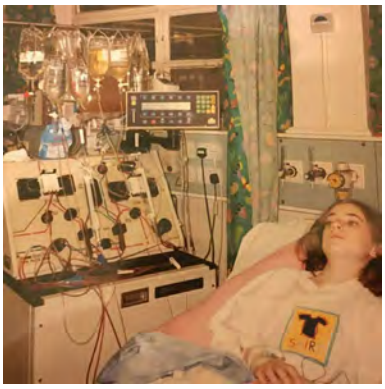
Paul's story

Paul Vallois, 49, received a life-changing kidney donation from his mother in 2011. Diagnosed with IgAN that year after experiencing tiredness, migraines and dangerously high blood pressure, Paul experienced a rapid decline in kidney function. Despite being a fit, healthy father of two young children, he found himself facing emergency dialysis and the need for an urgent transplant.

His mother stepped forward as a living donor. After only two days on dialysis, Paul received her kidney in September 2011. The transplant was a success and Paul's energy levels quickly returned. He resumed an active life, engaging in activities such as rock climbing and running, and even completed the London Marathon in 2019 to raise money for Kidney Research UK. Each year, Paul marks his 'kidneyversary' with flowers and a card for his mum, a heartfelt celebration of the second chance she gave him.

Today, Paul's kidney function is stable, though he acknowledges that the transplant is not a cure. He continues to monitor his health and knows he may need another transplant in the future. In the meantime, he lives life fully and encourages others to consider live donation and to stay alert for early signs of kidney disease.

Kathryn's story



Kathryn Croker was 13 when she was rushed to Great Ormond Street Hospital with stomach pain, joint swelling and a rash. Blood tests revealed signs of kidney failure. She was diagnosed with IgAV, a rare disease that damages small blood vessels, including those in the kidneys.

Within five months, Kathryn's kidneys failed. She began immediate dialysis and plasma exchange. At the time, there was no social media nor mobile phones, and being from a small village, she lost touch with her friends during her yearlong absence from school. High-dose steroids led to weight gain and body changes that affected her confidence.

Her kidney function eventually improved to around 40%, but she continued to struggle with fatigue and the side effects of treatment. She completed education and graduated from university, but her health continued to decline. In her 20s, she reduced her working hours due to extreme tiredness. Eventually, dialysis became necessary, but it took a heavy toll on her body. She experienced dangerously high blood pressure, and the treatment made her so unwell that she could not tolerate the four-hour sessions.

In 2013, at age 25, Kathryn received a kidney transplant from her father. The transplant restored stability, allowing her to regain some normality in her daily life. Today, her kidney continues to function, although she manages her health with ongoing medications and regular follow-up care.

Clinical perspective

Professor James Fotheringham, Consultant Nephrologist, and Dr Louise Oni, Consultant Paediatric Nephrologist

IgAN accounts for about 6% of all people with kidney disease.[134] Based on clinical trials, people with IgAN lose around 5 ml/min of kidney function per year.[134] Real-world data suggest many progress to kidney failure within 10–15 years.[126]

Liam's and Paul's stories illustrate how the absence of clear symptoms can complicate the diagnosis of IgAN.[135,136] Liam was diagnosed during a routine army medical, but the disease ultimately derailed his career plans and led to kidney failure and a transplant. Paul had non-specific symptoms like tiredness and migraines. But his condition was more advanced. He was admitted with very high blood pressure and diagnosed in hospital, needing emergency dialysis and eventually a transplant. Both men are now living well post-transplant, but their experiences highlight the unpredictability of the disease. The fact that IgAN can recur even after transplant[134,137] adds to the anxiety for people living with IgAN. Until recently, to treat IgAN we were repurposing existing treatments for other diseases, using generic kidney disease treatments or using steroid therapies in an untargeted fashion. We genuinely believe we are now getting a first glimpse of therapies that modify the underlying disease progress in a specific, targeted way.[135,136] The next critical step is ensuring early diagnosis and access to these new treatment pathways.

IgAV, formerly known as Henoch-Schönlein purpura, is a similar condition to IgAN. It usually affects children, often appearing after a simple cough, cold or stomach bug.[138–140] The faulty IgA protein causes inflammation not only in the kidneys but also in small blood vessels elsewhere, resulting in rashes, joint pain and gastrointestinal problems.[138–140]

Kathryn's case is a prime example of the seriousness of IgAV. She was 13 when her kidneys failed, requiring dialysis and plasma exchange within months of diagnosis. She is now thriving after receiving a transplant from her dad, but what is deeply concerning is that treatment options for children like her have barely changed in over two decades. While IgAN has over 100 clinical trials underway,[59,60] IgAV has only a handful globally,[141] which highlights how treatments for adults often become available faster than those for children. The conditions are so biologically similar that therapies could benefit both; however, we must do more to bridge the gap in paediatric research.

Financial, social and emotional impacts of rare kidney diseases

Despite the substantial impact of rare kidney diseases on those living with them, their carers and the NHS, robust economic data specific to each disease remain scarce. Most available information is derived from CKD data, which do not clearly outline the financial impact or resource use for each rare condition.

Because most rare kidney diseases lack targeted treatment, many people require dialysis or time-consuming medication regimens, which can be disruptive to daily life. For children and young adults, even minor absences from school can severely impact their educational attainment. Children on dialysis may miss more than 100 days of school a year.[133] Lower attendance is strongly associated with poorer attainment at both key stage 2 (ages 7–11), where pupils with higher absence rates score lower in primary school assessments, and key stage 4 (ages 14–16). At key stage 4, persistently absent pupils (defined as missing 10% or more of school sessions) are significantly less likely to achieve good GCSE passes in English and Maths.[142] Beyond the academic impact, each additional day of school missed between Years 7 and 11 is associated with an average lifetime earnings loss of approximately £750,[142] with those missing a year potentially losing up to £100,000 in lifetime earnings.[143] Persistently absent students are three times more likely to receive benefits Persistently absent students are three times more likely to receive benefits by age 28 and have about a 60% lower chance of sustained employment.[142]

Dialysis schedules and the physical toll of treatment likely contribute to these lower educational and employment outcomes. A UK survey of adolescents with kidney failure found that 78% of transplant recipients were in full-time employment compared with just 20% of those on dialysis.[144] Many of those diagnosed as children reported lasting impacts on education and job prospects. Broader national data confirm this pattern: average earnings for people with CKD fall sharply within months of diagnosis (about £293 per month) and remain lower for at least five years.[145] Average earnings fell by approximately £14,700 over five years after diagnosis. People with CKD are also 16% more likely to require benefits and are less likely to be in sustained employment within four years of diagnosis.[145]

Beyond the physical and financial impact, rare kidney diseases take a heavy emotional toll. Data from systematic reviews and international initiatives report concerning levels of anxiety, depression and suicidal thoughts among people living with these conditions and their caregivers.[146–149] Caregivers, mothers especially, often reduce hours or stop working, adding financial pressure.[148] Children with CKD struggle with body image, bullying, social isolation, restrictions on activities and psychological trauma from repeated medical interventions.[146,149] Many parents worry their children will never form lasting relationships or live independently.[146,149] Despite these profound needs, access to appropriate psychological support is often limited, especially in rural areas.[146,149]

Rare kidney diseases create a huge and wide-ranging impact on individuals, their families and the NHS. All these problems together – such as frequent hospital visits, high treatment costs, interrupted schooling and fewer job opportunities – make it much harder for people to live full lives and exacerbate existing inequities.

Summary and recommendations

Diagnostic challenges

Rare kidney diseases may mimic common conditions and are often asymptomatic at early stages, leading to delayed diagnoses or misdiagnoses. Delays in diagnosis can lead to further disease progression, and misdiagnoses can lead to inappropriate treatment, leaving **patients with worse outcomes and closer to kidney failure**.

Recommendation 1: Ensure sufficient and equitable access to diagnostics, including genetic testing

Recommendation 2: Develop targeted screening programmes for people at known risk of rare kidney diseases

Recommendation 3: Enhance education and support for kidney specialists and primary care providers to improve early recognition of potential rare kidney diseases and ensure timely, appropriate referrals for further evaluation and diagnosis

Fifteen-year-old Morven lives with cystinosis. Diagnosed in infancy, they now follow a strict, round-the-clock medication schedule, including through the night and at school, sometimes leaving them feeling socially isolated.

Healthcare challenges

NHS care pathways are not well adapted to the needs of people with rare kidney diseases, particularly children. Renal clinical networks and paediatric renal services improve coordination but lack national consistency, and there is inadequate support for transition from paediatric to adult care. In addition, regional access to specialist care is inconsistent. **Rare kidney disease care should be prioritised as the UK Government develops new policy to replace the current UK Rare Diseases Framework and ensure it is implemented across the four nations, to manage the disproportionate impact of rare kidney disease.**

"As more children with rare kidney diseases are now surviving into adulthood, nephrologists treating adults are increasingly encountering conditions they may not have seen before. Historically, many of these were considered childhood diseases. To keep pace with this shift, adult nephrologists need additional training and support to recognise and manage rare kidney diseases that may be unfamiliar to them."

Dr Ben Reynolds, Consultant Paediatric Nephrologist

Recommendation 4: Strengthen interconnection and collaboration between specialist centres and other health settings, e.g. via renal networks

Recommendation 5: Improve integration and continuity of care between paediatric and adult services, including increased support for transitional care and adolescent mental health services, and secure additional training for adult nephrologists on paediatric kidney diseases

Access to new medicines

The limited availability of disease-specific treatments means many people with rare kidney diseases rely on supportive or generalised treatments with considerable side effects. There is some hope, however, with a number of clinical trials ongoing for new targeted medicines. It is essential that regulatory and reimbursement pathways for medicines are optimised for rare diseases to **ensure patients receive access to new treatments as soon as possible**:

Recommendation 6: Ensure coordination between regulatory and reimbursement evidence needs so that rare disease studies are appropriately designed and inclusive across age groups

Recommendation 7: Include people with rare kidney diseases in research studies and clinical trials so that future treatments for mixed causes of CKD are also understood in rare diseases

> Gareth's kidneys were slowly deteriorating, but we embraced life together… We held on to the thought that advances in medicine could bring better therapies, or even a cure, in his lifetime." Due to a lack of treatment options for his ADPKD beyond hypertension control, he required heart and kidney transplants. He died aged 50.
>
> **Caroline Prodger, wife of Gareth**

Clinical trials

Enrolment in a clinical trial offers participants early access to new medicines and can boost UK R&D investment. However, in a recent review of 13 clinical trials for rare kidney diseases, 40% did not have a UK site, **meaning UK patients and the UK economy are not benefiting as much as they could be**. Furthermore, the exclusion of children from rare kidney disease trials means they are unable to benefit from new therapies at the same time as adults.

Recommendation 8: Support UK participation in regional and international clinical trials to increase UK patient access and representation

Recommendation 9: Address inequities around the exclusion of children from clinical trials where this is safe and appropriate

Recommendation 10: Offer everyone with a rare kidney disease the opportunity to participate in a clinical trial or research for their disease

Enhance RaDaR

RaDaR is a world-leading patient registry and an invaluable resource for the study of rare kidney diseases in the UK. However, enrolment into the database is time-consuming, and there is no government funding to support doing so. This means **we lack an accurate picture of the true prevalences of rare kidney diseases in the UK, and the data we do have are geographically skewed. Furthermore, patients who might benefit from participating in research are missing out**.

Recommendation 11: Increase capacity to enrol patients earlier, growing the cohort over time, to facilitate and expedite recruitment for clinical trials

Recommendation 12: Increase communication and education within the rare kidney disease community in the UK to foster trust and information exchange between patients and clinicians, and within RaDaR and beyond

Kathryn Croker was 13 when she was diagnosed with IgA vasculitis. Within five months, Kathryn's kidneys failed. She missed an entire year of school and, now in her twenties, works reduced hours due to ongoing symptoms and extreme tiredness.

Lack of economic evidence

Primary research into the economic impact of rare kidney diseases is lacking. A systematic review of the economic literature focused on rare kidney diseases published over the last ten years found 33 studies globally, but **none covered the UK**.

Recommendation 13: Demonstrate the economic value of delaying the progression of rare kidney diseases — either by reporting these conditions separately in large studies or conducting specific ones, e.g. through expansion and provision of access to the underlying dataset to researchers of the 2025 Office for National Statistics report: impact of health conditions requiring hospitalisation on earnings[145]

Recommendation 14: Ensure rare kidney diseases are accurately captured in administrative data, which inform cost analyses

Rare kidney diseases are a complex and underaddressed challenge within the UK healthcare system. While individually uncommon, their collective burden on patients, families and the NHS is substantial, ranging from delayed diagnosis and fragmented care to limited treatment options and minimal economic data. Patient experiences throughout this report highlight the personal and systemic toll of these conditions. To meaningfully improve outcomes, a coordinated response is needed that combines earlier detection, integrated and equitable care pathways, investment in targeted therapies and clinical research, and improved national data infrastructure. Implementing the recommendations outlined here is critical to ensuring that people with rare kidney diseases are no longer overlooked in policy, research and care delivery.

Acronyms

ADPKD	autosomal dominant polycystic kidney disease
AKI	acute kidney injury
AMKD	APOL1-mediated kidney disease
ANCA	anti-neutrophil cytoplasmic antibody
APOL1	apolipoprotein L1
APRIL	a proliferation-inducing ligand
ARPKD	autosomal recessive polycystic kidney disease
aHUS	atypical hemolytic uremic syndrome
BAPN	British Association for Paediatric Nephrology
BlyS	B-lymphocyte stimulator
CAKUT	congenital anomalies of the kidney and urinary tract
CAR	chimeric antigen receptor
CKD	chronic kidney disease
CSA-AKI	cardiac surgery-associated acute kidney injury
eGFR	estimated glomerular filtration rate
FSGS	focal segmental glomerulosclerosis
FXR	farnesoid X receptor
GMB	anti-glomerular basement membrane disease (goodpasture syndrome)
HST	highly specialised technology
HTA	health technology assessment
IgA	immunoglobulin A
IgAN	IgA nephropathy
IgAV	IgA vasculitis

IgG	immunoglobulin G
LDHA	lactate dehydrogenase A
LN	lupus nephritis
MCNS	minimal change nephrotic syndrome
MGRS	monoclonal gammopathy of renal significance
MHRA	Medicines and Healthcare products Regulatory Agency
NC	nephropathic cystinosis
NHS	National Health Service
NICE	National Institute for Health and Care Excellence
NURTuRE	National Unified Renal Translational Research Enterprise
PICOTS	population, intervention, comparators, outcomes, timeframe, and study design
PRISMA	Preferred Reporting Items for Systematic Reviews and Meta-Analyses
PUV	posterior urethral valve
R&D	research and development
RaDaR	National Registry of Rare Kidney Diseases
RCT	Randomised clinical trial
RNAi	RNA interference
SMC	Scottish Medicines Consortium
STA	single technology appraisal
TLR	targeted literature review
UK	United Kingdom
UKKA	UK Kidney Association

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Appendix

Targeted literature review methodology

Targeted literature reviews (TLRs) were conducted to identify the most recent publicly available evidence on economic and epidemiological outcomes across four rare kidney diseases – ADPKD, cystinosis, CAKUT and IgAV/IgAN – for both the UK and Western Europe. The reviews covered studies published from 2015 to April 2025 for economic outcomes and from 2022 to April 2025 for epidemiological data.

Searches were conducted in MEDLINE and Embase via OVID using systematic, reproducible strategies aligned with SIGN guidelines and recommendations from HTA agencies (NICE, IQWiG, HAS). Search eligibility criteria followed the PICOTS framework based on Cochrane guidelines. The TLRs included real-world observational studies (prospective, retrospective, registry-based and surveys) and economic evaluations for cost and resource input data.

Screening was performed in two stages – title/abstract and full text – by a primary reviewer with quality checks. A PRISMA-adapted flow diagram summarised the selection process. Multiple publications from the same study were consolidated, and data were extracted using a standardised Excel template with quality assurance to ensure completeness and accuracy.

Appendix Table 1. Summary of select rare kidney diseases, prevalence rates and outcomes based on published analyses of RaDaR cohorts.[6,26]

Rare kidney disease	Prevalence (per 100,000 population)	Median age at diagnosis (IQR), years	Median time from diagnosis to kidney failure (95% CI), years	Median time from diagnosis to death (95% CI), years	10-year patient survival (95% CI)
All RaDaR	35.90 (35.46–36.34)	40.6 (23.7–57.1)	–	–	–
Cystinosis	0.20 (0.17–0.23)	1.9 (0.7–9.9)	14.7 (13.0–16.1)	40.8 (37.8–NE)	0.99 (0.94–1.00)
X-linked Alport- female	0.41 (0.37–0.46)	26.4 (7.6–40.2)	40.3 (23.8–52.7)	NE (44.4–NE)	0.99 (0.97–1.00)
X-linked Alport- male	0.53 (0.48–0.59)	19.5 (8.2–34.5)	15.1 (12.9–17.2)	NE (50.1–NE)	0.97 (0.94–0.99)
ADPKD	9.87 (9.64–10.10)	38.3 (26.7–49.8)	22.6 (21.9–23.8)	72.9 (63.4–NE)	0.97 (0.97–0.98)
ARPKD/NPHP	0.30 (0.26–0.34)	8.7 (0.1–29.9)	19.3 (11.8–26.7)	50.5 (NE–NE)	0.95 (0.90–0.98)
ADTKD	0.26 (0.23–0.30)	42.7 (27.8–54.5)	16.6 (10.4–27.6)	51.0 (34.8–NE)	0.95 (0.90–0.98)
ANCA-associated vasculitis	2.67 (2.56–2.79)	61.9 (50.4–70.5)	34.9 (27.9–60.4)	29.8 (25.1–36.0)	0.82 (0.80–0.84)
Anti-GBM disease	0.16 (0.13–0.19)	54.9 (36.0–64.2)	0.1 (0.1–0.3)	NE (28.0–NE)	0.83 (0.74–0.90)
TBMN	0.22 (0.19–0.26)	31.7 (18.1–45.5)	33.3 (25.9–NE)	–	1.00 (1.00–1.00)
Cystinuria	0.64 (0.58–0.70)	31.6 (18.6–48.5)	–	NE (53.8–NE)	0.97 (0.95–0.99)
Hyperoxaluria	0.16 (0.13–0.19)	18.4 (4.5–36.3)	NE (42.0–NE)	NE (30.3–NE)	0.97 (0.88–0.99)
HNF1B mutations	0.12 (0.10–0.15)	13.1 (2.1–34.5)	22.3 (16.4–NE)	..	0.98 (0.89–1.00)

Rare kidney disease	Prevalence (per 100,000 population)	Median age at diagnosis (IQR), years	Median time from diagnosis to kidney failure (95% CI), years	Median time from diagnosis to death (95% CI), years	10-year patient survival (95% CI)
Renal cancer inherited	0.16 (0.13–0.19)	42.5 (26.0–53.8)	–	NE (17.2–NE)	1.00 (1.00–1.00)
Tubulopathies	0.56 (0.51–0.62)	15.5 (6.9–41.9)	NE (12.8–NE)	–	1.00 (1.00–1.00)
Tuberous sclerosis complex	0.34 (0.30–0.38)	15.4 (2.4–31.6)	61.0 (NE–NE)	58.1 (58.1–NE)	1.00 (1.00–1.00)
aHUS	0.38 (0.34–0.43)	19.9 (3.4–40.0)	21.7 (17.5–26.6)	NE (36.2–NE)	0.94 (0.89–0.97)
SSNS/MCD	2.29 (2.18–2.40)	16.8 (4.1–46.5)	54.3 (52.9–NE)	NE (54.1–NE)	0.97 (0.96–0.98)
SRNS/FSGS	1.96 (1.86–2.06)	27.7 (7.0–50.6)	16.5 (14.1–19.0)	–	0.94 (0.93–0.95)
IgA nephropathy	5.26 (5.10–5.43)	40.4 (29.5–51.9)	10.7 (10.1–11.7)	–	0.95 (0.94–0.96)
Membranous nephropathy	2.85 (2.73–2.98)	56.8 (45.4–67.2)	28.6 (22.4–32.3)	42.1 (31.3–48.0)	0.87 (0.85–0.89)
MGRS	0.20 (0.17–0.23)	62.7 (51.8–72.1)	8.2 (4.9–11.9)	16.0 (13.7–19.6)	0.78 (0.66–0.86)
MPGN/C3GN	1.29 (1.21–1.38)	34.1 (15.2–55.6)	17.6 (15.5–22.1)	49.2 (42.5–NE)	0.91 (0.88–0.92)
Retroperitoneal fibrosis	0.15 (0.13–0.19)	56.9 (51.5–66.8)	NE (20.5–NE)	22.0 (16.3–NE)	0.84 (0.75–0.91)
STEC HUS	0.23 (0.20–0.27)	3.4 (1.7–7.0)	22.9 (17.6–NE)	–	0.99 (0.94–1.00)
Other vasculitides	2.72 (2.60–2.84)	52.2 (24.0–66.6)	28.0 (25.4–35.1)	41.8 (34.9–NE)	0.85 (0.83–0.88)

Appendix Table 2. Select clinical trials for rare kidney diseases

Therapy name	Disease	Phase	Eligible age range	Mechanism of action	Sponsor	UK status / availability
Iptacopan	IgAN	Phase 3	18–100 years	Complement factor B inhibitor	Novartis	Phase 3 trial ongoing in UK; FDA approved for C3G
Atacicept	IgAN	Phase 3	≥18 years	Dual inhibitor of BLyS and APRIL, targets root immunological cause	Vera Therapeutics	Phase 3 ongoing, UK sites included
Zigakibart	IgAN	Phase 3	≥ 18 years	Anti-APRIL monoclonal antibody; reduces pathogenic IgA production	Novartis	Multiple UK sites, Phase 3 ongoing
Atrasentan	IgAN	Phase 2	Adults	Endothelin receptor antagonist; reduces proteinuria	Novartis	Multiple UK sites, Phase 2 complete
Sparsentan	IgAN, FSGS, IgAV, Alport syndrome	Phase 2	≥1 to <18 years	Dual endothelin angiotensin receptor antagonist	Vifor Pharma / Travere Therapeutics	NICE recommended for IgAN in adults; Phase 2 study enrolling
Farabursen	ADPKD	Phase 1b	18–55 years	miR-17 inhibitor; targets genetic drivers of cyst growth	Regulus Therapeutics	Only in US; Phase 3 trial planned Q3 2025
Inaxaplin	AMKD	Phase 3	≥10 years	Small molecule inhibitor of APOL1; targets underlying genetic cause; oral therapy. Phase IIa showed 47.6% reduction in proteinuria	Vertex Pharmaceuticals	Phase 3 global trial ongoing; includes adolescents; UK sites expected
Ravulizumab	CSA-AKI in CKD, IgAN, aHUS	Phase 3	18–90 years (CSA-AKI); adults (IgAN, aHUS)	Long-acting complement C5 inhibitor; prevents terminal complement activation	Alexion Pharmaceuticals	Phase 3 trials for CSA-AKI, IgAN and aHUS ongoing in UK and globally
Imlifidase	Goodpasture syndrome	Phase 3	≥18 years	IgG-degrading enzyme, enables rapid clearance of pathogenic antibodies	Hansa Biopharma	Pivotal Phase 3 ongoing, UK sites included
ABO-101	Hyperoxaluria	Phase 2/3	6–64 years	Gene therapy, AAV-mediated delivery for glycolate oxidase deficiency	Arbor Biotechnologies	FDA orphan drug; not yet in UK
Nedosiran	Hyperoxaluria	Phase 2	≥ 6 years	RNAi therapy targeting LDHA to reduce oxalate production	Dicerna Pharmaceuticals	Not yet approved in UK
Vonafexor	Alport syndrome	Phase 2	16–55 years	FXR agonist; anti-fibrotic and anti-inflammatory	Enyo Pharma	Phase 2 ongoing, global sites
DFT383	Nephropathic cystinosis	Phase 1/2	2–5 years	Potentially curative, gene-editing approach	Novartis	Only in US

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