GLOBAL PATIENT ALLIANCE for KIDNEY HEALTH

NOVEMBER 2025

OUTSIZED IMPACT:

Why Identifying the Cause of Rare Kidney Disease Matters

A Policy Brief from the Medical Advisory Council of the Global Patient Alliance for Kidney Health (GloPAKH)

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The Global Patient Alliance for Kidney Health's Medical Advisory Council lends clinical perspective to the organization's advocacy programming and helps identify unmet needs that can be addressed through heightened awareness and policy solutions.

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GLOBAL PATIENT ALLIANCE for KIDNEY HEALTH

The Global Patient Alliance for Kidney Health envisions health care systems that treat chronic kidney disease as a public health priority. By ensuring at-risk patients can access comprehensive screening and early treatment, disease progression can be slowed, and mortality prevented.

GLOBAL PATIENT ALLIANCE for KIDNEY HEALTH

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KEY POINTS

Chronic kidney disease is a major global health crisis that ranks among the top 10 causes of death worldwide.

Rare kidney diseases represent a subgroup of chronic kidney disease that disproportionately affects children and young adults.

If rare kidney diseases are detected early and managed optimally, progression to kidney failure, which requires lifelong dialysis or kidney transplantation, can be avoided or delayed in most cases.

A variety of treatments can slow the progress of rare kidney diseases, and new medications are now available in many countries, with more in development.

The young age of this population, combined with new treatment options, makes it more important than ever to identify, diagnose, and treat these patients as early as possible to allow them to lead productive lives and avoid costly and life-altering kidney replacement therapies.

Policymakers should prioritize rare kidney diseases along with chronic kidney disease and other non-communicable diseases in policies and budgets.

RARE KIDNEY DISEASES

A Critical Subset of Chronic Kidney Disease

Chronic kidney disease is an urgent global health problem that affects more than 10% of the world's population, accounting for more than 2.6 million deaths annually.[1,2] Early detection and treatment would alleviate human suffering and reduce the major economic impact of chronic kidney disease, which is on track to reach direct costs of $407 billion globally by 2027.[3]

Much of the world's chronic kidney disease burden is associated with aging and common cardiometabolic health conditions such as diabetes, obesity, cardiovascular disease, and hypertension. Although less frequent, inherited genetic mutations, aberrant immune responses, and environmental exposures are also important causes or risk factors.[4,5]

Rare diseases are defined by the European Union and many other countries as those affecting <1 per 2,000 people.[6] More than 500 kidney diseases fall into this category.[7] Several additional types of chronic kidney disease are more prevalent but share important features with their rarer counterparts. These include polycystic kidney disease and IgA nephropathy, both of which affect children as well as adults, and are not caused by the aforementioned common cardiometabolic conditions. As a result, these conditions are frequently considered rare kidney diseases, and are included here.

Causes of Rare Kidney Diseases

Rare kidney diseases have different underlying causes, many of which are poorly understood.

GENETICS

Many rare kidney diseases are caused by or associated with genetic alterations that can take several forms.

- **Monogenic (also called single gene or Mendelian):** Monogenic alterations involve inherited mutations in a single gene, which cause up to 50% of pediatric and 10% of adult kidney disease.[8] These conditions often affect more than one person in a single family, placing a particularly high burden on those affected and limiting the opportunities for living donor transplants.
- **More common genetic variants that increase risk in certain groups:** Some genetic changes, such as in the APOL1 gene in people with African ancestry, make kidney disease more likely, especially in people with other common conditions such as high blood pressure or certain infections. Groups in which these gene variants are more common have a higher burden of kidney disease.

To complicate matters, genetic alterations often interact with non-genetic factors such as infections, immune responses, environmental exposures, and lifestyle to influence the risk of chronic kidney disease in ways that are not fully understood.

IMMUNE-RELATED DYSFUNCTION

Many rare kidney diseases are characterized by immune-related dysfunction, including conditions in which the body's immune system mistakenly attacks its own proteins—so-called autoimmune disease.[9] The reason that the immune system attacks the kidneys in certain people is poorly understood. Autoimmune kidney diseases can be associated with genetic risk factors, environmental exposures, and/or systemic diseases such as lupus or other infections. Some are more common in certain groups of people.

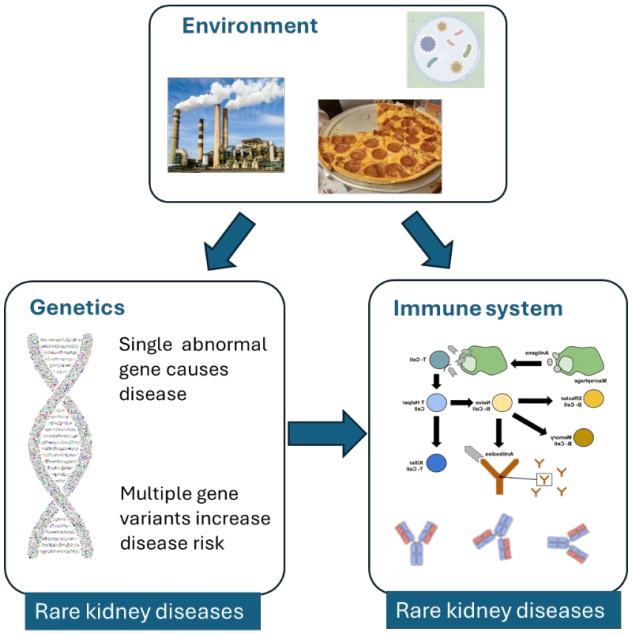


FIGURE 1

UNKNOWN CAUSES

Not infrequently, the cause of chronic kidney disease among patients undergoing kidney dialysis is reported as unknown. It is likely that many of these cases are caused by established rare kidney diseases that have not been diagnosed due to a lack of testing or uncertain diagnosis.[10] Uncertain diagnosis can be due to co-existing features of more than one disease, further complicating matters.

Given the variety of causes of rare kidney diseases, their overlapping nature, and the frequency of cases with unknown causes, there is no single widely accepted classification scheme. Instead, rare kidney diseases are often categorized based on a combination of underlying cause and the part of the kidney affected.[11] A comprehensive list of individual rare kidney diseases can be found on the European Rare Kidney Disease Reference Network website.[12]

Given the variety of causes of rare kidney diseases, their overlapping nature, and the frequency of cases with unknown causes, there is no single widely accepted classification scheme.

IMPORTANCE OF DIAGNOSING RARE KIDNEY DISEASES

Over the past decade, major advances have been made in diagnosing and treating chronic kidney disease that have the potential to slow and even prevent progressive loss of kidney function that can lead to kidney failure.[4]

Research is uncovering new genes and improving our understanding of immune-related and environmental contributions. Knowing the cause of a patient's kidney disease provides important information about prognosis[13] and allows individualized patient and family management. Patients themselves report improved understanding of their diagnosis and a change in their medications following a biopsy (examination of a small sample of their kidney tissue).[14] Precise diagnosis is increasingly important as more treatments become available that target underlying biological mechanisms and pathways, including specific genes and proteins.

"An important reason to pursue diagnosis is so that patients have information about their predicted disease course over the long term. Expecting a progression to dialysis in 5 years is very different than 20 years. As new treatments become available, of course diagnosis becomes even more important."

DR. DANIEL GALE

In addition to giving patients and families a prognosis and enabling personalized treatment, diagnosis of rare kidney diseases is particularly important due to the disproportionate effect on children and young adults, the prolonged burden on patients and caregivers, and the disproportionate contribution of rare kidney diseases to kidney failure.

Disproportionate Effect on Children and Young Adults

Rare kidney diseases disproportionately affect children and young adults. Indeed, the vast majority of chronic kidney disease cases in the pediatric population are due to rare kidney diseases.[15,16]

Children are also over-represented among patients receiving kidney dialysis or kidney transplantation. One European registry study reported that nearly 58% of children receiving these treatments had a rare disease.[17]

Unfortunately, diagnosis of rare kidney diseases is often delayed. Patients may spend years visiting multiple healthcare providers before receiving an accurate diagnosis.[18] Such delays cause psychological anguish for patients and their loved ones and delay treatment.[19] In most patients, the disease is not diagnosed until 50% of kidney function has been lost or injuries sustained that lead to dramatically reduced kidney function.[20,21] Description of actual diagnostic journeys from patients and parents are available at the CompCure and Rare Kidney Disease Foundation websites.

"It can be difficult to diagnose rare kidney diseases, as the early symptoms tend to be unspecific or very particular. As a result, many patients and parents must navigate a difficult diagnostic journey, where you need to be a bit lucky, or very assertive and resourceful, to find a center that has the right level of competence required to specifically diagnose certain rare kidney diseases."

MS. MARIANNE SILKJÆR NIELSEN

Prolonged Burden on Patients and Caregivers

Rare kidney diseases place an undue and lifelong burden on patients, caregivers, and families. These conditions can inflict a major negative impact on the daily lives and productivity of both patients and caregivers.[22,23] Patients experience anxiety and depression and are less likely to participate in social activities.[22] Their ability to engage in work or school is also commonly affected, with reduced productivity, increased absenteeism, reduced career advancement, and early job discontinuation.

Caregivers also experience anxiety and depression, in addition to work interference. Caregiver burden increases as the patient's health and functional independence decline.[24] Female gender, lower caregiver and patient income, longer time providing daily care, and longer duration of caregiving are also associated with increased caregiver burden.[24] Furthermore, parents often feel guilty about passing genetically linked diseases to their children.[22]

For both patients and caregivers, the financial stress caused by missed work and reduced work productivity is compounded by the direct costs of healthcare co-payments, medications and diagnostics, medical devices, and transportation to and from medical appointments.[22] Patients often delay or forego diagnostic tests and treatment because they can't afford them.

Disproportionate Cause of Kidney Failure

Rare kidney disease is a disproportionate cause of kidney failure. A large registry study from the United Kingdom found that patients with rare kidney diseases have higher 5-year rates of kidney failure but higher survival than other patients with chronic kidney disease (stage 3 or higher) and are over-represented among patients requiring kidney replacement therapy.[13] Another large registry study in Europe found that diabetes and hypertension accounted for only 37% of people initiating kidney replacement therapy, while rare kidney diseases accounted for about 40%.[25]

Kidney failure is associated with a drastically reduced life expectancy and years of productive life lost, even among patients undergoing treatment.[20] Notably, screening efforts focusing on diabetes mellitus will miss most of the patients with rare kidney diseases.

Although chronic kidney disease associated with diabetes and hypertension can progress to kidney failure, older patients with these conditions often have competing risk factors such as cardiovascular disease that cause death before they require dialysis or kidney transplantation.[26]

> "The disproportionate contribution of rare kidney diseases to kidney failure has not really changed over the past 30 years. We are seeing this in an analysis of our Daily Outcomes and Practice Patterns Study in dialysis patients around the world."
>
> **DR. ROBERTO PECOITS-FILHO**

SOCIOECONOMIC AND HEALTH BENEFITS OF IMPROVING DIAGNOSIS OF RARE KIDNEY DISEASES

Improving diagnosis of rare kidney diseases can help reduce the staggering burden these conditions place on patients, caregivers, healthcare systems, economies, and the environment.

Improve Patient Health

A diagnosis makes an enormous difference to patients and families, who often spend months or years consulting different healthcare providers undergoing test after test. Once a diagnosis is made, patients may be matched with disease-specific treatments that target the underlying cause of their condition.[27] Even in cases where no disease-specific therapies are available, accurate diagnosis can facilitate a treatment plan that may help slow disease progression, reduce complications, and avoid unnecessary interventions. The humanistic benefits of treatments that allow children and adults to pursue their interests instead of spending their days undergoing kidney dialysis are difficult to estimate.

A diagnosis also enables patients to participate in clinical trials evaluating new treatments and registries designed to collect data to improve disease understanding and treatment. Finally, patients and families with gene-based kidney diseases can benefit from genetic counseling and family planning.

"The cost of disability due to rare kidney disease in young people is huge and it's not getting better. We urgently need strategies to address this worldwide."

DR. PATRICK MARK

Alleviate Caregiver Burden

As noted in the previous section, rare kidney diseases place a substantial burden on caregivers. Given that caregiver burden increases as the patient's health and functional independence decline,[24] obtaining treatment that improves patient health may help reduce caregiver burden. Patients who receive effective treatment, particularly early in the disease course, can live with less disability and may be able to delay or even avoid kidney replacement therapy.[28,29] This may reduce the time needed from caregivers, leading to less work disruption and increased ability to engage in social activities. Transportation time and costs are also major concerns for caregivers,[22,24] and early diagnosis and treatment of rare kidney diseases that allow patients to avoid or delay kidney dialysis are also likely to alleviate caregiver burden.

Reduce Impact on Health Systems and the Environment

The progression of rare kidney diseases is overwhelming, not only for patients, caregivers, and families, but also for healthcare systems and economies. Advanced kidney disease is associated with more outpatient visits, pharmacy claims, and higher total costs than earlier-stage disease.[30] This is evidenced by a study of patients with IgA nephropathy in which costs were significantly higher in patients with high-risk (≥1g/d) versus low-risk (<1g/d) proteinuria: $3732 vs. $1457 per patient per month.[30]

Once rare kidney diseases have progressed to kidney failure, the costs of kidney replacement therapy—dialysis or kidney transplantation—continue throughout patients' lives. The global costs of dialysis alone for patients with rare kidney diseases have been estimated at more than $73 billion annually.[19]

Earlier diagnosis and treatment of rare kidney diseases would reduce these costs and are particularly important given the evolving treatment landscape.

Kidney dialysis is associated with significant carbon emissions due to use of water, single-use plastic, energy, and medical waste.

In Canada alone it has been estimated that three-times weekly hemodialysis of 20,000 patients over one year produces carbon emissions equal to circumnavigating the earth in an average-sized car 7785 times.[31] In the United Kingdom, hemodialysis requires 94,000 liters of water and 3000 kWh of electricity per patient per year.[32]

Peritoneal dialysis has been less thoroughly studied, but it requires transporting plastic-packed fluid across and between countries and is therefore expected to have a substantial environmental impact.[33]

A reduction in the number of patients undergoing kidney dialysis would help minimize the impact of this treatment on the environment.

TABLE 1. ESTIMATED GLOBAL ANNUAL COST OF DIALYSIS FOR RARE KIDNEY DISEASES[19]

DISEASE	ESTIMATED GLOBAL COST
ADPKD	$57.2 billion
Membranous nephropathy	$7.1 billion
IgA nephropathy	$6.5 billion
Alport syndrome	$1.3 billion
C3 glomerulopathy	$644 million
Focal and segmental glomerulosclerosis	$451 million
Atypical hemolytic uremic syndrome	$419 million
Goodpasture syndrome	$0.4 million

ADPKD=autosomal dominant polycystic kidney disease

MAKING THE DIAGNOSIS OF RARE KIDNEY DISEASES

Chronic kidney disease can be screened for using urinalysis and blood testing, but such tests are often insufficient to establish a cause for rare kidney diseases, particularly in children.[34] Instead, diagnosis of rare kidney disease often requires the use of kidney biopsies, genetic tests, and/or imaging.

A careful clinical history and detailed physical exam are also important to establish a family history and assess other symptoms that are characteristic of specific rare kidney diseases.

- **Kidney biopsies:** Tests in which tiny samples of the kidney are obtained, usually by inserting a needle through the skin, and examined under a special microscope to evaluate the appearance of kidney tissue and cells.
- **Genetic tests:** Tests in which a tissue sample from the inner cheek, saliva, blood, or other tissue is examined for alterations to genes or chromosomes.
- **Imaging:** Visualization of the kidneys using ultrasound, computerized tomography (CT), and magnetic resonance imaging (MRI) to assess kidney size, presence of cysts, blood flow, and other information.
- **Evidence of metabolic abnormality:** For certain metabolic diseases (eg, Fabry disease), blood testing may assess enzyme activity or accumulation of specific metabolites.
- **Clinical history and physical exam:** A thorough clinical history should seek to establish family history of kidney disease, age at onset, and other relevant details. Symptoms in other body systems, detected by clinical history or physical exams, also inform diagnosis and may themselves require treatment.

The most recent KDIGO (Kidney Disease Improving Global Outcomes) guidelines emphasize the importance of establishing a cause for chronic kidney disease using the resources available.[4]

It is never too late to diagnose the cause of kidney disease. For example, appropriate diagnosis of patients who have undergone a kidney transplant may lead to interventions that prevent disease recurrence in the transplanted organ.[36]

"Although the KDIGO and other guidelines recognize a family history of kidney disease as a reason to screen for chronic kidney disease,[4,35] this is rarely implemented in the clinic and, when done, it is implemented suboptimally. Family history should be assessed periodically (e.g., yearly), as it may evolve over time as the first family member requires kidney replacement therapy, the only event serious enough to be commented on by family members."

DR. ALBERTO ORTIZ

TREATMENT STRATEGIES FOR RARE KIDNEY DISEASES

General Treatments

For many rare kidney diseases, treatment involves a comprehensive strategy with a multidisciplinary care team,[4] that may include the following general treatments.

- **Lifestyle changes:** active lifestyle and a diet with more fruits and vegetables and lower ultraprocessed foods
- **Medications that target generic drivers of disease progression:** renin-angiotensin system inhibitors, sodium–glucose cotransporter-2 inhibitors (SGLT-2is), nonsteroidal mineralocorticoid receptor antagonists (NSMRAs), glucagon-like peptide-1 (GLP-1) receptor agonists, and other antiproteinuric agents that might be effective in slowing progression in some rare diseases
- **Management of chronic kidney disease-related complications**

Despite the above interventions, in most rare kidney diseases, outcomes have remained poor for many years, with the exceptions being those diseases for which highly effective, specific disease-targeted treatments have become available (see next section).

For monogenic diseases, cascade testing of relatives, predictive testing (to allow earlier protective interventions and inform life, transplant, and reproductive decisions) and reproductive interventions can be important in addressing needs of those affected.

> "We cannot generalize care of patients with rare kidney disease. If we personalize and give precise care, we will have improved patient outcomes."
>
> **DR. VICTORINE NZANA**

Additionally, given the many different causes of rare kidney diseases, treatments can vary considerably. Personalized care, including matching treatment to the diagnosis, is critical for those with rare kidney diseases given the potential for targeted treatment, as well as the complexity and variability of disease complications.

Disease complications vary widely based on the diagnosis—some rare kidney diseases are caused by genetic conditions that affect multiple body systems, such as vision and hearing. In contrast, others are more likely to affect cardiovascular function, cause blood clots, or damage the brain. A precise diagnosis can help healthcare providers evaluate and treat suspected complications.

"Diagnosis leads to specific actions that can be missed in patients who are undiagnosed. There is a big variability in management strategies."

DR. ROBERTO PECOITS-FILHO

Specific Treatments

A number of new medications for rare kidney diseases are now available in various countries, with many more in development.[37] Advances in basic science research and improved understanding of rare kidney diseases causes and mechanisms have led to medications that target blood vessels, fluid balance in the kidneys, metabolic defects, and various immune system pathways. Advances in genomics and gene delivery have resulted in several gene-based treatment strategies, some of which are at advanced stages of preclinical or even clinical testing.[38] Treating patients with appropriate medications can lead to enormous benefits that include delaying kidney failure.[13]

"A specific diagnosis is required in order to get a targeted therapy."

MS. MARIANNE SILKJÆR NIELSEN

Examples of treatments specifically for rare kidney disease include eculizumab[39] and ravulizumab[40] for atypical hemolytic uremia syndrome and paroxysmal nocturnal hemoglobinuria; tolvaptan[41] for autosomal dominant polycystic kidney disease; iptacopan[42] and pegcetacoplan[43] for C3 glomerulopathy; atrasentan,[44] budesonide[45], iptacopan[42], and sparsentan[46] for IgA nephropathy; lumasiran[47] and nedosiran[48] for primary hyperoxaluria type 1; agalsidase-alfa[49,50], agalsidase-beta[50,51], pegunigalsidase-alfa[52,53], and migalastat[54,55] for Fabry disease; and voclosporin[56] and belimumab[57] for lupus nephritis.

Unfortunately, most of the medications specifically for rare kidney diseases are available only in a few regions worldwide. Part of the challenge is that, although rare kidney diseases as a whole affect a large population, each individual disease affects a much smaller group, making it difficult to enroll enough people in the clinical trials needed for medication approval. The small populations also reduce financial incentives for companies to market their medications. In this regard, consolidation of health data for specific rare diseases may make it easier, cheaper, and less risky to develop therapies for rare kidney diseases.

However, for many countries, the medications are simply unaffordable. This has led some countries to pass legislation designed to promote drug development for rare diseases.[58] This is important because use of these medications has real potential to improve lives and limit resources expended on kidney dialysis and transplantation.

"Currently, there are few genetic kidney conditions that can be targeted with specific and highly effective therapies that are needed to prevent (rather than simply delay) kidney failure, although this may change if effective genetic therapies can be developed for kidney diseases."

DR. DANIEL GALE

Even if there is no specific treatment for a given rare kidney disease, knowing the cause may allow healthcare providers to avoid prescribing useless medications that compromise patient safety, such as immune suppressants for metabolic rare kidney diseases.[59]

Additionally, clinical trials of new treatments for rare kidney diseases are ongoing in various countries worldwide. By participating in clinical trials, patients can often access medications that would not be available to them otherwise, while also participating in data collection that may eventually enable the medications to reach more patients.

"Regulatory action in the United States and Europe has improved the development of new medications for rare kidney diseases. However, there is still a focus on higher-income countries, which is a barrier that must be addressed."

DR. NAVDEEP TANGRI

"India is a part of several global trials for rare kidney diseases, including IgA and membranous nephropathy. Some of the trials provide for post-trial drug access, which is welcome because many of the drugs are not approved quickly in India."

PROF. MANISHA SAHAY

"Many rare kidney diseases are poorly understood and insufficiently studied, which leads to weak guideline recommendations, varying standards of care, and insufficient evidence to meet regulatory requirements."

MS. MARIANNE SILKJAER NIELSEN

A Case of Childhood Diagnosis Without Available Therapy

A 6-year-old female child was referred with deformities of lower and upper limbs (knock knees) since 2 years of age. A wrist X-ray showed cupping and fraying of metaphyses.

The laboratory evaluation showed a variety of abnormalities (eg, proximal renal tubular acidosis). Slit-lamp examination of the eye showed refractile intracellular crystals, which are pathognomonic for cystinosis in the setting of Fanconi syndrome; thus, the child was diagnosed with cystinosis, a rare genetic disorder caused by the intracellular accumulation of the amino acid cysteine. Renal failure in Franconi syndrome occurs by adolescence. The child was placed on supportive therapy. Specific treatment consists of cysteamine tablets and eye drops (even after kidney transplantation), although these treatments are not available freely in low- and middle-income countries.

Case contributed by Dr. Manisha Sahay

A Case of Alport Syndrome Caught Early and Effectively Managed

Mr. CR, a 20-year old adult, presented with macroscopic hematuria—the third episode that he had been managing with rehydration. He reported blurred vision and progressive hearing loss. His physical exam revealed grade 2 hypertension, proteinuria, and normal kidney function. He was referred to the ophthalmologist for suspected anterior lenticonus, which was confirmed. As relevant history, his older brother presented with similar symptoms and was on maintenance hemodialysis; he was also found to have anterior lenticonus, and both boys were diagnosed with X-linked Alport syndrome. Presently, Mr. CR is on angiotensin converting enzyme inhibitors, with well controlled blood pressure, minimal proteinuria, and acceptable glomerular filtration rate.

Case study contributed by Dr. Victorine Nzana

POLICY OPPORTUNITIES

Diagnosis and treatment of rare kidney diseases are evolving rapidly, and the time is right for policy changes designed to help patients, caregivers, economies, and the environment.

Policymakers should take the following actions to help realize the benefits of diagnosing rare kidney diseases:

Implement general screening for chronic kidney disease as a first step.

Prioritize rare kidney diseases along with chronic kidney disease and other non-communicable diseases in policies and budgets.

Facilitate international rare kidney disease research and evidence generation to improve disease understanding across countries, ethnicities, clusters, etc., which will inform guidelines, decision-making and also de-risk research and development programs, thereby attracting investments in new therapies and diagnostic tools across rare kidney diseases.

Establish centers of excellence and multidisciplinary teams to provide access to genetic and other advanced tests, experienced pathology teams, evidence-based standard of care, and multidisciplinary teams to help address complications and maximize patient functioning.

Develop and implement clinical guidelines to help standardize best-practices care.

Develop disease-specific care pathways based on current evidence.

Promote healthcare provider education so that they can conduct the appropriate tests and referrals to obtain accurate diagnoses.

Promote general public awareness through education programs for all classes of society.

> "We need to educate healthcare providers so that they can get tests to patients, especially outside specialty centers."
>
> **DR. PATRICK MARK**

Through these actions, we can begin to address the multifaceted burden of rare kidney diseases and work toward alleviating the lifelong suffering of patients and families worldwide.

APPENDIX

Current Approach to Screening and Diagnosis Across Different Regions

"The key decision where I work with regard to glomerular disease is whether or not to perform a kidney biopsy. The benefits of making a diagnosis early in the disease are evolving over time and being able predict the disease course over time is an important consideration."

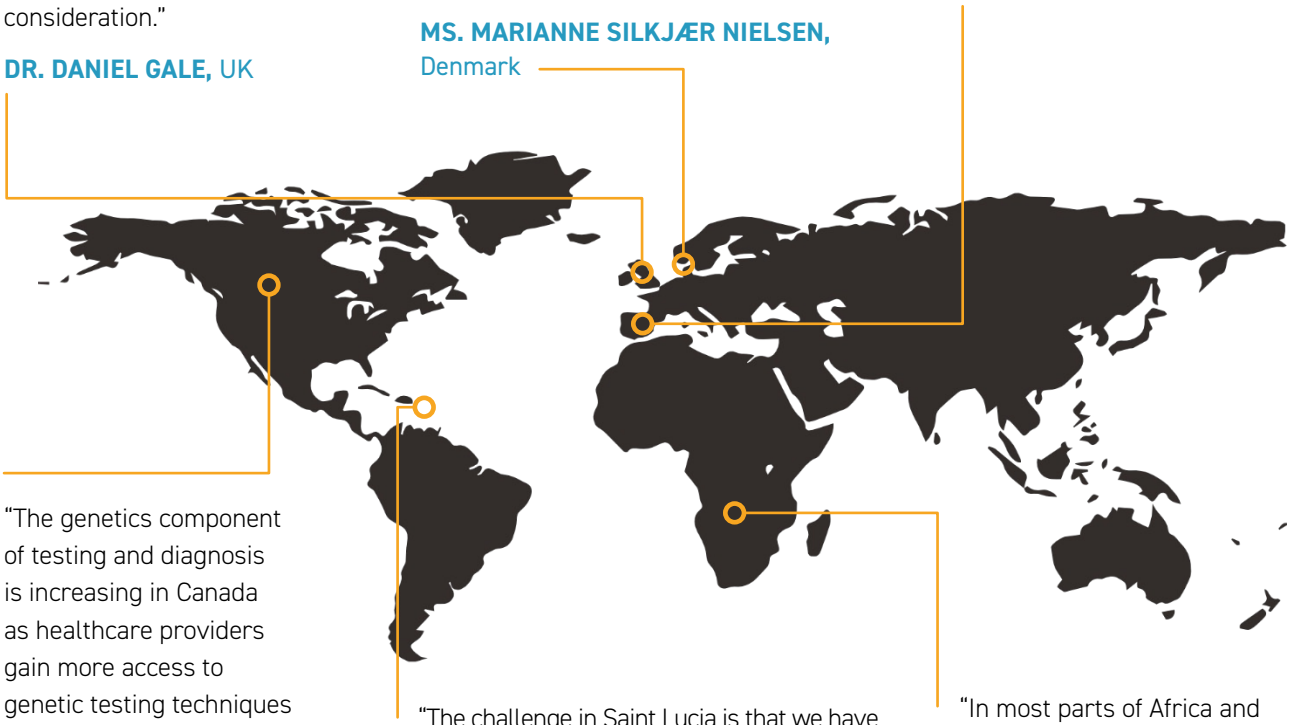
DR. DANIEL GALE, UK

"With the exception of Japan, routine screening for kidney disease is not done consistently as part of a regular health check program. It may be beneficial at least to screen certain high-risk populations such as children born prematurely. Although such screening would be easy and cheap, it is not routinely done, even in high-income countries."

MS. MARIANNE SILKJÆR NIELSEN, Denmark

"The importance of screening is demonstrated by a recent study in Spain, which found that 1 in 4 people under the age of 45 years who undergo kidney replacement therapy due to unknown or unclear causes have genetic findings that could explain their kidney failure."[10]

DR. ALBERTO ORTIZ, Spain



"The genetics component of testing and diagnosis is increasing in Canada as healthcare providers gain more access to genetic testing techniques and experience with interpretation of these findings."

DR. NAVDEEP TANGRI, Canada

"The challenge in Saint Lucia is that we have many young patients on dialysis without a definitive diagnosis. We essentially put the cause of end stage renal disease as unknown because they present too late for biopsy and genetic testing is not available. A few years ago we tested for Fabry disease and everyone came back negative. So, we are in the position where we assume that some of our patients may have a rare disease but we can't say so definitively."

DR. MERLE CLARKE, St. Lucia (Caribbean)

"In most parts of Africa and especially in sub-Saharan Africa, it's very complicated to diagnose rare kidney disease. Most areas don't have genetic testing and biopsies are rarely done. We do have imaging for cystic diseases but, overall, it's a bit challenging to diagnose rare kidney disease."

DR. VICTORINE NZANA, Cameroon

Example Organizations that Promote Clinical Trials in Rare Kidney Diseases

Belgian pediatric clinical trial network: promotes clinical research and facilitates high-quality trials

CompCure: unites multiple stakeholders and develops evidence in two rare kidney diseases, which supports research, development, guidelines, etc.

Connect for children network (C4C): helps optimize pediatric trial facilitation

Cure Glomerulopathy Network: a team of medical research professionals who study glomerular disease in children and adults.

European Rare Kidney Disease Reference Network (ERKNet): information on rare kidney disease populations

IGA Nephropathy Foundation: patient-centric organization focused on finding a cure for IgA nephropathy

International Society of Glomerular Disease: professional society working for accurate, early diagnosis and suitable treatments to preserve kidney health

Neph Cure: empowers patients to take charge of their health and lead the revolution in research

Nephrotic Syndrome Study Network (NEPTUNE): seeks to match patients to therapies that can effectively treat their rare kidney disease

PKD Foundation: funds research, advocates for patients, and supports a community for all impacted by polycystic kidney disease

Rare Disease Action Network / American Kidney Fund: advocates for legislation and policies; works to improve awareness of rare diseases

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