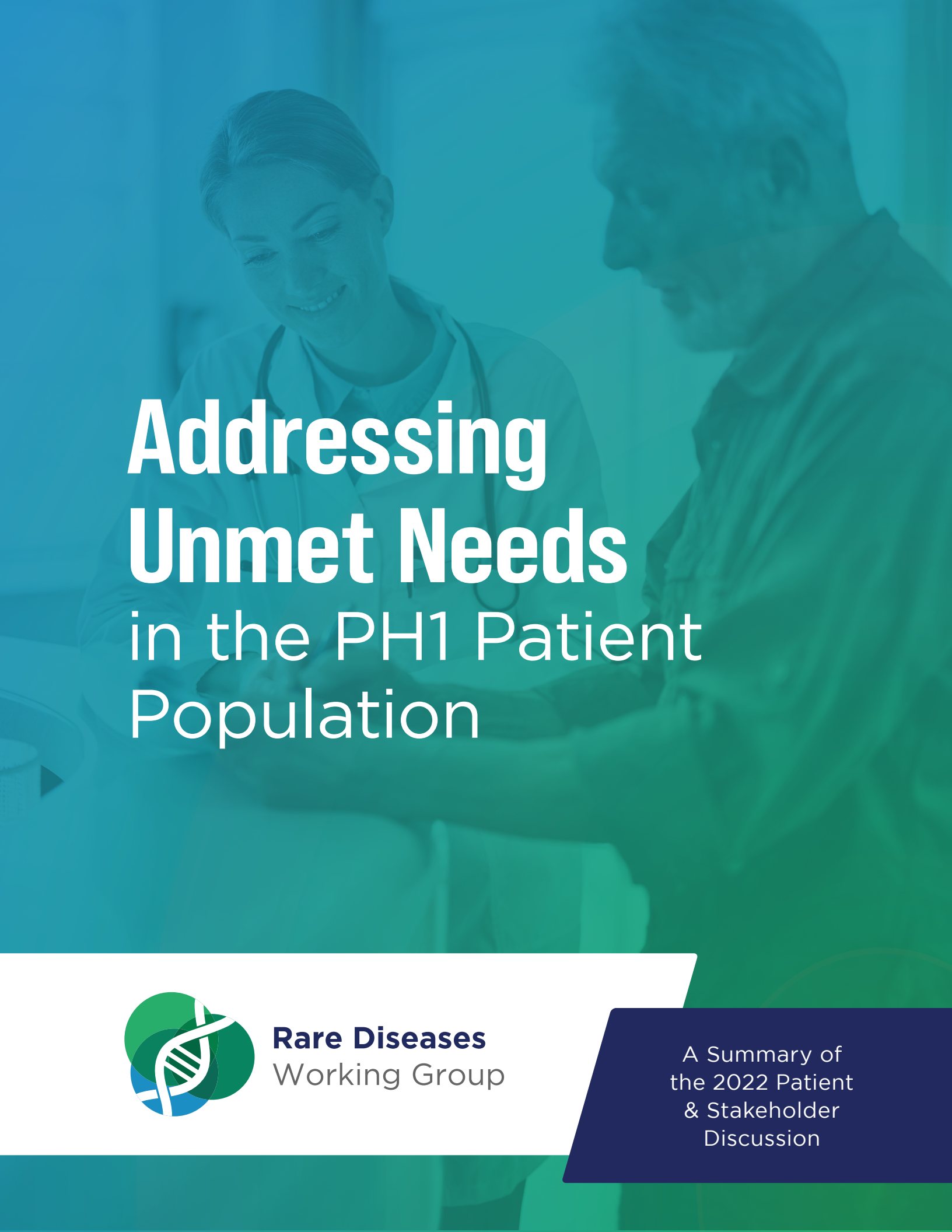

# Addressing Unmet Needs in the PH1 Patient Population

Rare Diseases Working Group

A Summary of the 2022 Patient & Stakeholder Discussion

# Overview

**On 13 July 2022, the Alliance for Patient Access' Rare Diseases Working Group** hosted a virtual discussion on primary hyperoxaluria type 1. Patients and stakeholders representing physicians and patients across the PH1 spectrum discussed patients' experiences and identified ways to better support patients with this ultra-rare disease.

## Primary Hyperoxaluria Type 1

Primary hyperoxaluria type 1 is a rare genetic condition characterized by too much oxalate in a patient's urine. Oxalate is a chemical that naturally occurs in the body and is also found in some foods. When oxalate builds up, it can lead to painful or life-threatening symptoms, leading to kidney failure in some cases. The hallmark symptom, though, is frequent and numerous kidney stones that often require surgical intervention.

The liver normally produces proteins that help prevent the buildup of oxalate. But for patients with primary hyperoxaluria type 1, the liver doesn't make enough of the enzyme that reduces oxalate levels, leading to the development of kidney stones.

Patients are born with the genetic disease, but it sometimes goes undetected for decades. This can create a challenge for adults who are undiagnosed. In most cases, however, patients are diagnosed at a young age because they develop symptoms like kidney stones, which are otherwise rare among young people. These symptoms are tell-tale signs for physicians that the child may need testing for other conditions.

> "PH1 is like a ticking time bomb where the first indication could be kidney failure."
>
> - Kim Hollander, Oxalosis & Hyperoxaluria Foundation

# Identifying Community Needs

The condition's rarity and symptoms both present unique challenges. Recognition that multiple kidney stones may be a sign of this rare disease is still lacking. The discussion with patients and stakeholders focused on identifying patients' unmet needs.

## Identifying Adult PH1 Patients

While many patients are diagnosed as children, misdiagnoses and a lack of awareness mean that many adults go under or undiagnosed.

"Patients with PH1 often receive a misdiagnosis more than once or go years undiagnosed, as too often rare diseases are off the radar to health care providers. It is imperative that health care providers look behind what is causing kidney stones in children or recurrent kidney stones in adults," Kim Hollander explained.

Without proper treatments, adults with PH1 may suffer preventable damage. Increased awareness among the public and health professionals is needed to ensure correct and timely diagnoses.

## Fragmentation Between Medical Specialties

Because PH1 is caused by a deficiency in the liver and results in frequent kidney stones, patients may receive care from or be diagnosed by a variety of specialists. Many patients are treated by nephrologists, but some patients may see a urologist or even a hepatologist. This can create a challenge for patients, as they navigate the linkage between specialists and seek continuity of care.

The rarity of PH1 creates an additional layer of complexity to this challenge. Patients with frequent kidney stones are unlikely to ask their providers about PH1 and specialists may not know enough about the disease to test stones for oxalate levels. Increased communication between providers is needed to improve diagnoses and treatment outcomes.

"For about 6-8 years, I was traveling once a month for testing. I was losing a lot of time at home with my family and the activities I loved doing most."

- Mayah B., PH1 Patient Advocate

## Smooth Care Transitions

Primary hyperoxaluria type 1 is often diagnosed when patients are young. As patient attendees described, they were fortunate enough to, once diagnosed, work with a team of providers who understood their rare disease and oversaw their treatment throughout childhood and adolescence.

Transitioning from pediatric care to adult care as they grow older can be difficult. Patients explained that moving from a specialist who knows your condition and background and starting over with a new specialist, who may not understand the rare condition as well, can be disorienting and frustrating.

## Public Awareness

While patients are often unaware of their condition before receiving a diagnosis, many providers — even those who specialize in kidney or liver disorders — are also unfamiliar, given PH1's rarity.

Stakeholders and patients discussed how a lack of awareness can lead to a long wait to diagnosis, misdiagnoses and ineffective — even harmful — treatment plans. Providers need to be equipped with all the information necessary to support their kidney stone patients.

## Specialist Care

Patients and stakeholders explained that seeing a specialist who understands the condition makes a significant difference when it comes to diagnosis and optimal care.

But for many patients, finding a provider who can properly diagnose and treat the condition can be a complicated process.

## Patient Connection & Education

The rarity of primary hyperoxaluria type 1 also makes it difficult for patients to connect with others and learn more about their condition. The educational materials that do exist are often targeted more toward medical professionals than patients.

Rigorous treatment often requires patients to be away from school, work or home for extended periods of time. Those disruptions can impact their social life and undermine relationships. Meeting participants agreed that creating a way for patients to connect with each other could provide meaningful social benefit.

## Access to Genetic Testing & Counseling

Primary hyperoxaluria type 1 is a genetic condition. A recessive gene causes the enzyme deficiency responsible for the rare condition. If a child receives the recessive gene from both parents, they have primary hyperoxaluria type 1.

When patients are diagnosed, genetic testing can provide answers for both patients and their families. Patients can find comfort in a confirmed diagnosis, and genetic testing may inform family members if they also have the disease or are carriers.

But genetic testing comes with a great deal of emotional pressure. For a disease like primary hyperoxaluria type 1, the rare condition's complicated nature may intimidate patients. Improving education on the value of genetic testing, and supporting coverage that allows for both testing and counseling, will make a difference for patients and their families, providing closure and certainty.

"Patients need to understand they deserve a definitive diagnosis and it's okay to get a second opinion. We should also educate about the value of genetic testing."

- Michael Spigler, American Kidney Fund

# Potential Next Steps

Patients and stakeholders also discussed what can be done to overcome the challenges patients may face.

## Improve Partnerships Among Advocacy Organizations

Advocacy organizations can help connect patients with the right resources, such as educational materials or specialists. By playing to their strengths and referring patients to other organizations with the most ideal connections, advocacy organizations can collaborate to ensure that patients receive the best, most personalized care for their needs. For example, allied patient advocacy groups could collaborate on a consensus paper, host a joint event for kidney stone patients or collaborate on other initiatives.

## Promote Innovative Treatment Options

Primary hyperoxaluria type 1 patients now have an approved therapy for the first time. Ensuring timely access to different treatment options is critical to determine the best course of action. Groups could collaborate to highlight new treatment options and address access barriers.

## Increase Public Awareness

Promoting primary hyperoxaluria type 1 and finding ways to educate patients and providers about this disease should also be a top priority. Increased awareness may improve the likelihood of receiving a diagnosis or a referral to a specialist. Creation of new educational materials, cross-promotion of resources on social media or a joint educational campaign are several possible methods for this work.

## Conclusions

Primary hyperoxaluria type 1 can be dangerous and life-threatening for patients. Its painful symptoms and often arduous diagnostic odyssey can introduce both physical and emotional challenges. But finding ways to meet the needs of this patient community through advocacy and collaboration can strengthen patients' resolve and improve their quality of life.

## Participants

AfPA Alliance for Patient Access

Oxalosis & Hyperoxaluria Foundation
STEPPING STONES TO A CURE

AAKP American Association of Kidney Patients

American Kidney Fund®
FIGHTING ON ALL FRONTS

American Urological Association

GLI Global Liver Institute

National Kidney Foundation®