LIVING WITH PH1
A guide for caregivers

This guide was created to help both you and your child integrate primary hyperoxaluria type 1 (PH) management into your daily lives.

living with PH1

Developed and funded by Alnylam® Pharmaceuticals

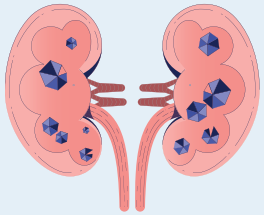
Covering the basics

Whether your child was diagnosed yesterday, or years ago, it is important to understand the basics of PH1.

PH1 is a rare, inherited, recessive disease. **Inherited** means that it is passed down within families. **Recessive** means both your mother and your father have passed the mutated (changed) gene that causes PH1 down to you. With PH1, the liver makes too much of a waste product called **oxalate**. In a healthy liver, oxalate is present only in small amounts, is not used by the body, and is removed by the kidneys. However, with PH1, the liver makes too much oxalate, which can cause **crystals** to form in the kidneys, causing progressive damage. **Progressive** means the damage is continuously happening to the kidneys and can be getting worse over time, even if the symptoms sometimes aren't noticeable.

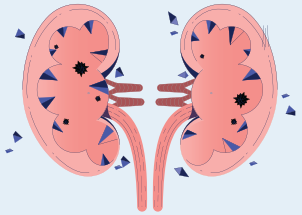
Crystals cause damage in different ways

1. Joining together to create kidney stones



2. Collecting in the kidneys and causing damage (nephrocalcinosis).

   Over time, as kidneys are damaged, oxalate can spread and form crystals in other parts of the body (systemic oxalosis).



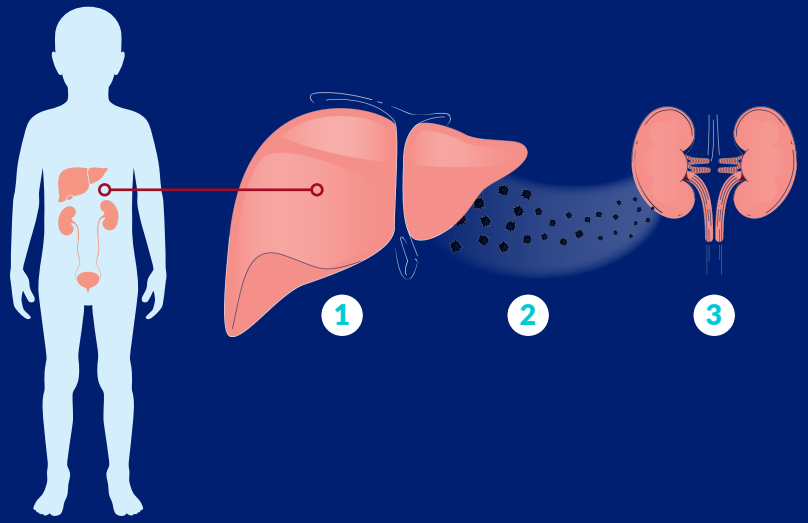
Could someone else in your family have PH1?

Because of the inherited nature of PH1, if your child has a PH1 diagnosis confirmed by a genetic test, it is important that family members, especially siblings, should be tested as well. **Speak to your doctor about getting genetic tests for your family members.**

Give your child a look inside PH1

PH1 starts in the liver but can hurt the kidneys

1. The liver makes **too much oxalate** the body can't use.
2. The body **sends the oxalate to the kidneys** to try to get rid of it.
3. But oxalate **can turn into crystals**, which can build up and damage the kidneys. Over time, the kidneys stop working properly.



Looking for more?

Watch *How PH1 Happens* at **LivingwithPH1.ca** and find out more about the science behind your child's condition.

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Working with your healthcare team

While managing PH1 can feel overwhelming at times, you are **not alone**. Your child's healthcare team will help you to navigate the challenges ahead. Depending on how your child's PH1 is progressing, your nephrologist will create a customised care plan.

MY CHILD'S PH1 DOCTOR:

NAME:

CONTACT INFO:

Monitoring your child's health

There are several different ways your care team may track PH1 and check for kidney damage. The frequency of the tests and procedures used will depend on your child's care plan. Below are some terms you may hear your care team use when discussing how they monitor your child's health.

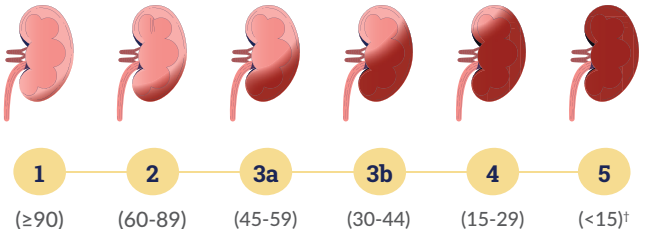
TERM	DEFINITION
KIDNEY FUNCTION BLOOD TEST	Test that checks how well the kidneys are filtering waste from blood
KIDNEY TRANSPLANT	A kidney transplant is a surgical operation where a donor kidney is taken from a person without PH1 and is transplanted into a person with PH1, who may have had kidney damage cause by their PH1
KIDNEY ULTRASOUND	Imaging that detects and monitors any kidney stones that may form
LITHOTRIPSY	Non-invasive procedure to break up kidney stones*
LIVER TRANSPLANT	A liver transplant is a surgical operation where a donor liver is taken from a person without PH1 and is transplanted into a person with PH1. A liver transplant therefore stops oxalate being over produced in the liver.
URETERO AND RENAL ENDOSCOPY	Minimally-invasive procedure to remove/break up stones*
URINARY OXALATE TEST	Test that keeps track of oxalate levels in urine

* A urologist will select a procedure based on the size/amount of stone(s).

Checking on kidney function

Doctors measure kidney function by checking the **estimated glomerular filtration rate (eGFR)**. Your child's eGFR number is based on a blood test for creatinine, a waste product in the blood.

Because PH1 can lead to **chronic kidney disease (CKD)**, it's important to check eGFR levels and kidney function regularly. The stages of CKD range from low risk of kidney damage (stage 1) to kidney failure (stage 5).



† These values correspond to how many milliliters per minute the kidney is filtering and is adjusted for body area.

Talking to your child about PH1

Teaching your child the value of their care plan can help them stay motivated.
Try sharing these points to help them understand why managing PH1 is important:

Hyperhydrating and taking medicine

- Drinking lots of water helps dilute oxalate — that means making it weaker.
- It's not always easy drinking so much water — especially if you're not feeling thirsty. Your healthcare professional team will advise how much water is right for you.
- Ask your child — what are some good ways I can help you?
- Taking vitamin B6 may help your body make less oxalate. Your healthcare professional team will advise how much vitamin B6 is right for you
- Taking alkali citrate may help lower the number of crystals that are made up of oxalate.

Taking care of body and mind

- You can help keep your body as strong as possible by eating healthy foods, playing outside with friends and getting a good night's sleep.
- It's okay to feel tired, angry, or sad about having PH1 sometimes. Come and talk to me, and I'll do whatever I can to help.

Going to dialysis

- When kidneys can't do their job anymore, they will need some extra help. There is a machine that can work like kidneys— it's called a dialysis machine.
- Dialysis will help get rid of the excess oxalate.
- While the dialysis machine does its job, you'll need to sit still and wait patiently. Are there any activities, games, or books you'd like to bring?

Going to the doctor

- Doctors play an important role in managing your PH1. Doing tests helps them know how to take care of you.
- Doctors can help if you get a stone or if your kidneys aren't strong enough. They can also answer any questions you may have about PH1. Is there anything you want to ask the doctor at our next visit?

Have a curious child?

PH1 of a Kind™ is an animated video series made just for kids living with PH1. Watch them with your child at **LivingwithPH1.ca**.

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Preparing for your child's journey with PH1

Staying on top of PH1 management can help to slow the damage to the kidneys, but too much oxalate can cause permanent damage. Eventually transplant surgery may be necessary if the PH1 has become too advanced. Researchers are actively working to develop additional ways to treat PH1.

Understanding transplant surgeries

A liver transplant is a surgical operation where a donor liver is taken from a person without PH1 and is transplanted into a person with PH1.

A liver transplant therefore stops oxalate being over produced in the liver. Most other management options cannot do this.

Because PH1 causes damage to the kidneys, both the liver and the kidneys may need to be replaced, either at the same time or during separate surgeries. This is called a dual liver-kidney transplant.

Transplant surgeries are major operations that require preparation, long-term follow-up, and lifelong medications. Talk to your healthcare professional team about developing a detailed plan to help you recover from your surgery.

Wherever your child is in their PH1 journey, you may have to teach others about their disease

Not everyone will be familiar with PH1, and this may include some members of the healthcare professional team. However, you can play an important role in educating others about the disease. Try coming up with a quick description of PH1 that you can share with others. You can use the space below to jot it down.

IF ASKED ABOUT PH1, I'LL SAY:

Looking for more?

To learn about other ways you can help your child manage PH1, go to **LivingwithPH1.ca**.

Helping your child succeed

A solid routine can help your child stay on top of management, so they can stay involved with the things they love.

Tips for appointment prep

- Ask clinic or hospital staff if there's anything your child needs to do beforehand
- Record any symptoms, even if they seem unrelated to PH1
- Track how much your child drinks and urinates over the course of a day
- Make a list of all medications, vitamins, or other supplements your child takes
- Write down questions to ask your doctor

Tips for sticking to the care plan

- Reward healthy habits and celebrate small wins
- Set reminders on your child's phone/watch to remind them to drink water
- Designate a special water bottle or bracelet, as a reminder to drink enough water
- Anticipate frequent bathroom breaks
- Plan ahead for events that could disturb or interrupt the usual care plan (holidays, school trips, etc.)

Tips for reading the signs

Because your child might have trouble describing how they feel, you may notice symptoms before they can articulate them. Keep in mind that **your child may not feel any of these symptoms**, and still have a stone or kidney failure. If you think your child is experiencing these symptoms, you may consider contacting your nephrologist or urologist.

SIGNS OF A KIDNEY STONE	SIGNS OF KIDNEY FAILURE
• Pain in side or back • Blood in urine • Frequent need to urinate • Difficulty urinating	• Decreased urine or no urine output • Loss of appetite, nausea, vomiting • Pale skin colour • Swelling of hands and feet • Extreme fatigue and weakness

Looking for more?

To learn about other ways you can help your child manage PH1, go to **LivingwithPH1.ca**.

Finding strength & support

As a caregiver, it's important to remember your own wellbeing.

Practice self-care

You are the strongest part of your child's support system. It takes time, energy, and perseverance, so don't forget to take care of yourself. That means setting aside time to eat healthy, stay active, unwind, and pay attention to your own psychological wellbeing.

Expand your circle of support

It's important that those who your child interacts with regularly understand how PH1 can affect your child's day-to-day—like babysitters, teachers, and sports coaches. For instance, telling your child's coach about PH1 may be helpful so they can help ensure that your child is drinking enough water and they can appreciate the need to take frequent bathroom breaks.

There's a form you can download, fill out, and share with others to inform them about your child's PH1.

You can find it at LivingwithPH1.ca.

By communicating your child's unique needs to other people, you can expand the network of knowledgeable adults looking out for your child's wellbeing.

Looking for more?

To hear stories from others living with PH1, go to **LivingwithPH1.ca**.

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Expand your circle of support

Healthcare professionals, advocacy groups, and other people living with or caring for someone with PH1 can be great resources for additional tips and guidance. Below are three resources that are just a search away.

Canadian Organization for Rare Disorders

CORD is Canada's national network for organizations representing all those with rare disorders. CORD provides a strong common voice to advocate for health policy and a healthcare system that works for those with rare disorders. CORD works with governments, researchers, clinicians and industry to promote research, diagnosis, treatment and services for all rare disorders in Canada.

RQMO
REGROUPEMENT QUÉBÉCOIS DES MALADIES ORPHELINES

RQMO provides rare disease patients and their families with Information, resources and support to help with managing rare disease. We connect people with knowledgeable physicians, researchers and clinics specialized in the disease, and official sources of information on medical management and treatment. We help patients find government and community resources to help with medical services; financial and legal help; respite and home care; and psychosocial support. We provide basic genetic counselling and answers to questions about genetic tests, possible risks, prenatal diagnosis, etc.

kidney FOUNDATION

About The Kidney Foundation of Canada

Excellent kidney health, optimal quality of life for those affected by kidney disease, and a cure. This vision has guided us to be a collaborative, inventive and focused leader in the development of programs, services, research opportunities and awareness campaigns that have had a positive impact on the millions of Canadians living with, or at risk of developing kidney disease. The Foundation's national research program has grown to become one of the most important sources of funding for scientists conducting kidney-related research. The Foundation is committed to providing education, information and support about kidneys and kidney disease.

Looking for more?

For more information, visit **LivingwithPH1.ca**.

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