

Go further.

Get to the root cause of early-onset or recurrent kidney stones with NovoDETECT™.

Easy, seamless genetic testing can facilitate earlier diagnosis and help determine your next steps



Actor portrayals.



Sponsored by Novo Nordisk, NovoDETECT™ genetic test kits are available through **Blueprint Genetics**, which also provides support at **(833) 472-2999**.

Make diagnosing easier



For pediatric patients <18 years of age with a single stone or anyone experiencing recurrent kidney stones (RKS):

- Diagnosing the underlying causes can be complicated by the rarity of associated hereditary conditions, wide clinical variability, and overlapping symptoms of many genetic kidney stone disorders^{1,2}
- Underlying causes of RKS often go undiagnosed, which can result in progressive damage to the kidneys, leading to chronic kidney disease (CKD) and eventually end-stage kidney disease (ESKD)¹⁻³
- Delays in diagnosis can be detrimental^{2,4}

Actor portrayals.

Key indicators of a genetic kidney stone disease, including primary hyperoxaluria (PH)^{2,3,5-8}:

- A single kidney stone in an infant or child
- RKS in adults
- Nephrocalcinosis
- Family history of kidney stones
- Elevated urine oxalate levels
- Advanced CKD with unknown cause
- Failure to thrive and ESKD in infants
- Signs of systemic oxalosis

In a study, **~15%** of patients with nephrolithiasis/nephrocalcinosis had a causative monogenic condition.⁹

Studies have shown that children with nephrolithiasis have as high as a **50% risk of recurrence within 3 years** following a kidney stone event.¹⁰

Could it be PH?

Early-onset kidney stones or RKS may be an indication of PH, a group of rare, genetic metabolic disorders caused by monogenic, biallelic mutations in *AGXT*, *GRHPR*, or *HOGA1*, resulting in elevated urinary oxalate and the formation of calcium oxalate crystals.^{6,8}

PH carries a significant patient burden, often requiring dialysis and dual liver/kidney or kidney transplant.¹¹

PH causes hepatic oxalate overproduction, which can lead to^{6,8}:



Nephrocalcinosis



Progressive kidney damage or ESKD



Systemic oxalate deposition

Early, accurate diagnosis and management of PH can help to slow disease progression.⁴

Visit [UncoveringPH.com](https://uncoveringph.com) to learn more.



The value of genetic testing

Gene-specific analysis can be pivotal for accurate diagnoses due to symptomatic overlap in the clinical presentation of patients with kidney stone diseases.¹²

Identifying disease-causing mutations can allow for patient management that may reduce recurrent symptoms or progression to ESKD.¹³

A definitive diagnosis of PH requires genetic testing.^{3,14} Visit the NovoDETECT™ genetic testing program at NovoDETECT.com to get started with uncovering the underlying genetic cause of early-onset kidney stones or RKS.

The answers and support you need



Sponsored by Novo Nordisk, **NovoDETECT™ offers no-charge genetic and PH-specific metabolite testing**—essential tools that can help identify the underlying cause of early-onset kidney stones or RKS. As a seamless end-to-end experience, NovoDETECT™ can provide clarity to help guide your next steps for patients.

No patients, healthcare professionals, or payers, including government payers, are billed for this program.

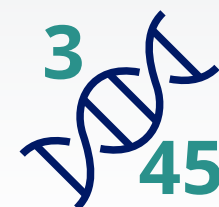


Actor portrayals.



Customer-centric services

- ✓ A **Blueprint Genetics** support team serves as a single point of contact for accessing, ordering, and processing tests through our **call center at (833) 472-2999**
- ✓ **Clinical Genomic Services (CGS)** consultants are also available to help you answer clinical questions, select the right test, and interpret laboratory results
- ✓ Pre- and post-result genetic counseling is available, which you can opt in to easily for your patient when ordering a test. Patients will be contacted by the genetic counseling support team to schedule an appointment



Comprehensive genetic testing

- ✓ Straightforward genetic testing for eligible patients through **Blueprint Genetics** to help increase diagnostic accuracy
- ✓ 3-gene PH panel tests for mutations in PH-associated genes: *AGXT* (PH1), *GRHPR* (PH2), and *HOGA1* (PH3)
- ✓ 45-gene nephrolithiasis panel tests for mutations in PH-associated genes as well as assessment of a multitude of other genes associated with kidney stone disease

Genetic testing may identify a variant of uncertain significance (VUS) in a patient for whom, at the time of interpretation, the pathogenicity is unknown.¹²



VUS Resolution Program in PH

- ✓ Working together with **Quest Diagnostics**, a PH urine metabolite assay is conducted to help resolve a VUS result in PH, including whether it is pathogenic or benign
- ✓ Offers segregation studies to investigate how PH was inherited and how it may impact family members
- ✓ Further investigates PH-associated VUS results, whether the result was reported by a different diagnostic testing company or through this program

Learn more at [NovoDETECT.com](https://www.novodetect.com) about what to do with PH VUS results.



Support is available at **(833) 472-2999**.

Get started with a no-charge **NovoDETECT™ test kit**.

Patient eligibility for genetic testing

For patients to be eligible for genetic testing through NovoDETECT™, they must live in the US or a US territory and **meet at least 1** of the following criteria.

Adult/pediatric



Family history of RKS and/or monogenic kidney stone disorders^a resulting in RKS



Advanced CKD of unknown etiology



Individuals with previous genetic testing with a VUS^b reported in *AGXT*, *GRHPR*, or *HOGA1*



Laboratory indication (urine/blood biochemistry or stone analysis composition) of monogenic disorders resulting in RKS (ie, elevated oxalate in urine, plasma, or oxalate within stone analysis)



Nephrocalcinosis

Pediatric



Children (<2 years old) with failure to thrive and impaired renal function



Kidney stones

- Adults (≥18 years of age) with history or presence of bilateral/multiple/RKS
- Pediatrics (<18 years of age) with history or presence of ≥1 kidney stone

^aIncludes hyperoxaluria, hypercalciuria, hyperphosphaturia, hypocitraturia, hyperuricosuria, and cystinuria.

^bWhere a genetic variant has been detected but pathogenicity is indeterminate.

Getting started

The NovoDETECT™ program makes it easy to place your order, follow progress, and review results.

Step 1



Order NovoDETECT™ genetic test for eligible patients

- Genetic tests can easily be ordered through either:
 - 1 A seamless and secure online portal, Nucleus, which can be accessed at NovoDETECT.com or
 - 2 A downloadable Test Requisition Form available via NovoDETECT.com that can be returned via email, fax, or mail
- When ordering, be sure to complete the necessary information pertaining to the selected panel and your patient
- Order 1 of 2 panels: **PH panel (3 gene) or nephrolithiasis panel including PH (45 gene)** with choice of blood **or** buccal test
 - Be sure to select the panel that is most appropriate for your patient
- When ordering, **you can immediately opt in to pre- and post-result genetic counseling** to help guide you and your patient through the testing process

Step 2



Receive test kit and collect sample

- Kits will contain the consent form that a patient needs to review, sign, and return with the sample for the test to move into analysis
- Buccal sample kits can be sent directly to your patient's home or your office for collection. Blood samples can be collected in your office or in the patient's home through **ExamOne**
- Kits contain all materials and prepaid shipping labels

Step 3



Review test results

- At any time, via the Nucleus portal, easily check the status of your order and review test results—often available within 6 weeks
- Discuss test results and next steps with your patient, including family testing for PH

NovoDETECT™ is not intended to and should not interfere in any way with a healthcare professional's or patient's independent judgment and choice in the treatment options for these diseases. Healthcare professionals and patients should always consider the full range of treatment options and select those most appropriate for the individual patient.



Support is available at **(833) 472-2999**.

Get started with a no-charge **NovoDETECT™ test kit**.

A strong foundation of support throughout the diagnostic journey



- Comprehensive genetic and metabolite testing to assist you in diagnosing your patients at high risk for underlying genetic cause of RKS
- Board-certified geneticists and genetic counselors to support you and your patients throughout the process
- PH VUS resolution program designed to determine the pathogenicity of poorly understood VUS
- Support for patients with biallelic mutations, including those with a VUS in a PH-associated gene
- Novo Nordisk is committed to helping the nephrolithiasis community identify the underlying causes of RKS and other kidney stone disorders through comprehensive, accurate diagnostic testing

Call NovoDETECT™ at (833) 472-2999 (Monday-Friday, 8 AM-8 PM EST) to speak with a **Blueprint Genetics** support team member for assistance throughout the diagnostic journey.

For more information, visit [NovoDETECT.com](https://www.novodetect.com).

No patient-identifiable information or raw sequence data will be shared outside of the program. Examples of de-identified patient data are clinical diagnosis, age range, sex, and genetic variants associated with kidney stone diseases. Contact information of the healthcare professional associated with the patient may also be shared as needed.

No samples or identifiable research data will be shared with third parties without express permission from the patient.

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Blueprint Genetics



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