NOVODETECT™
Driving change in the diagnostic journey

When a genetic report reveals a variant of uncertain significance (VUS) in PH-associated genes

Move closer toward certainty with NovoDETECT™

Confirming a diagnosis of primary hyperoxaluria (PH) requires definitive genetic results. For those with a VUS result in *AGXT, GRHPR,* or *HOGA1* genes, additional evaluation of metabolites related to oxalate can be beneficial in assessing enzyme activity.[1]

Sponsored by Novo Nordisk, the NovoDETECT™ PH VUS Resolution Program can help clarify a PH diagnosis

Committed to help provide answers and reduce the number of patients with inconclusive results in a PH gene

A urine metabolite assay that includes glycolate, glycerate, and 4-hydroxy-2-oxoglutarate (HOG) to specifically detect PH1, PH2, and PH3.[1] Positive or negative results may allow for VUS reclassification[2,3]

A segregation study examining gene distribution among family members can help determine family inheritance patterns, which can be informative for patients with PH-positive or VUS results[3,4]

Board-certified geneticists and genetic counselors to support healthcare professionals and their patients who have been opted in for genetic counseling

For more information, visit NovoDETECT.com.

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Identifying a VUS

If a VUS is identified through **Blueprint Genetics**[a] the NovoDETECT™ PH VUS Resolution Program can help clarify results.

If a patient received a VUS result from another lab's diagnostic panel, a new panel assessment will be conducted.

PH metabolite testing is provided at **no charge** to you, your patients, or their healthcare insurance.

Provide any patient medical events or family history not previously reported via the Nucleus portal.

A segregation study can help identify parental origins of PH variants (mutations) and determine if family members are carriers of those variants or are at risk for PH.[3,4]

Urine will be collected conveniently in the patient's home by an **ExamOne** home health technician and tested by **Quest Diagnostics**.

Tests are conducted by **Quest Diagnostics**. **Blueprint Genetics** evaluates results, including assessment of PH metabolite testing results, clinical symptoms, urine oxalate levels, family history, and genetic testing.

Laboratory results along with geneticist evaluation and recommendations are noted and made available within approximately 6 weeks.

Clinical Genomic Services (CGS) consultants are available to address clinical questions or help interpret the genetic testing report.

[a]Patients who receive a VUS result from **Blueprint Genetics** will automatically be enrolled in the NovoDETECT™ PH VUS Resolution Program.

Connect with a **Blueprint Genetics** support team member at **(833) 472-2999** (Monday-Friday 8 AM-8 PM EST).

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.

References: 1. Sas DJ, Harris PC, Milliner DS. Recent advances in the identification and management of inherited hyperoxalurias. *Urolithiasis.* 2019;47(1):79-89. **2.** Schlosser P, Li Y, Sekula P, et al. Genetic studies of urinary metabolites illuminate mechanisms of detoxification and excretion in humans. *Nat Genet.* 2020;52(2):167-176. **3.** Blueprint Genetics. VUS—the most maligned result in genetic testing. Accessed August 30, 2023. https://blueprintgenetics.com/resources/vus-the-most-maligned-result-in-genetic-testing/ **4.** Ferraro PM, D'Addessi A, Gambaro G. When to suspect a genetic disorder in a patient with renal stones, and why. *Nephrol Dial Transplant.* 2013;28(4):811-820.

Blueprint Genetics

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