MAYO CLINIC LABORATORIES

Test Definition: HYOX

Hyperoxaluria Panel, Random, Urine

Overview

Useful For

Distinguishing between primary and secondary hyperoxaluria

Distinguishing between primary hyperoxaluria types 1, 2, and 3

Genetics Test Information

Primary hyperoxalurias, classified into types 1, 2, and 3, are genetic disorders of oxalate metabolism characterized by increased urinary excretion of oxalic acid and kidney stone formation.

Secondary hyperoxaluria is an acquired condition resulting from either increased intake of dietary oxalate or altered intestinal oxalate absorption.

Testing Algorithm

For more information see Hyperoxaluria Diagnostic Algorithm.

Special Instructions

- Hyperoxaluria Diagnostic Algorithm

Highlights

A diagnostic workup in an individual with hyperoxaluria demonstrates increased concentration of oxalate in urinary metabolite screening. If glycolate, glycerate, or 4-hydroxy-2-oxoglutarate is present, a primary hyperoxaluria is indicated.

Each type of primary hyperoxaluria is distinguished from the others based on the urine profile.

Method Name

Gas Chromatography Mass Spectrometry (GC-MS)

NY State Available

Yes

Specimen

Specimen Type

Urine

Necessary Information

1. **Patient age is required.**
2. Biochemical Genetics Patient Information (T602) is recommended, but not required, to be filled out and sent with the

specimen to aid in the interpretation of test results.

Specimen Required

Supplies: Urine Tubes, 10 mL (T068)
Container/Tube: Plastic, 10-mL urine tube
Specimen Volume: 10 mL
Collection Instructions:
1. Collect a random urine specimen.
2. No preservative.
3. Immediately freeze specimen.

Forms

1. Biochemical Genetics Patient Information (T602)
2. If not ordering electronically, complete, print, and send 1 of the following forms with the specimen:
-Biochemical Genetics Test Request (T798)
-Renal Diagnostics Test Request (T830)

Specimen Minimum Volume

1.1 mL

Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Urine	Frozen (preferred)	90 days	
	Refrigerated	14 days	

Clinical & Interpretive

Clinical Information

Increased urinary oxalate frequently leads to kidney stone formation and kidney insufficiency. Identifying the cause of hyperoxaluria has important implications in therapy, management, and prognosis.

Hyperoxalurias are classified as primary and secondary. Primary hyperoxaluria is an inherited disorder of oxalate metabolism, while secondary hyperoxaluria is an acquired condition resulting from either increased intake of dietary oxalate or altered intestinal oxalate absorption. Primary hyperoxalurias are classified into types 1, 2, and 3.

Primary hyperoxaluria type 1 (PH1) is an autosomal recessive disorder resulting in a deficiency of peroxisomal alanine:glyoxylate aminotransferase (AGT) due to variants in the *AGXT* gene. It is characterized by increased urinary oxalic, glyoxylic, and glycolic acids. PH1 is the most common type with manifestations that include deposition of calcium oxalate in the kidneys (nephrolithiasis, nephrocalcinosis) and kidney failure. Calcium oxalate deposits can be further deposited in other tissues, such as the heart and eyes, and lead to a variety of additional symptoms. Age of onset is

variable with a small percentage of patients presenting in the first year of life with failure to thrive, nephrocalcinosis, and metabolic acidosis. Approximately half of affected individuals show manifestations of PH1 in late childhood or early adolescence, and the remainder present in adulthood with recurrent kidney stones. Some individuals with missense *AGXT* variants respond to supplemental pyridoxine therapy. Two RNA interference (RNAi) therapies for PH1 are also available, lumasiran and nedosiran. Additionally, liver transplantation can restore hepatic AGT enzyme activity.

Primary hyperoxaluria type 2 (PH2) is due to a defect in the *GRHPR* gene resulting in a deficiency of the enzyme hydroxypyruvate reductase. PH2 is inherited in an autosomal recessive manner and is identified by an increase in urinary oxalic and glyceric acids. Like PH1, PH2 is characterized by deposition of calcium oxalate in the kidneys (nephrolithiasis, nephrocalcinosis), and kidney failure. Most individuals have symptoms of PH2 during childhood, and it is thought that PH2 is less common than PH1. Treatment is focused on lowering calcium oxalate through daily fluid intake and inhibitors of calcium oxalate crystallization.

Primary hyperoxaluria type 3 (PH3), due to recessive variants in *HOGA1* (formerly *DHDPSL*), occurs in a small percentage of individuals with primary hyperoxaluria. *HOGA1* encodes a mitochondrial 4-hydroxy-2-oxoglutarate aldolase that catalyzes the 4th step in the hydroxyproline pathway. PH3 is characterized biochemically by increased urinary excretion of oxalate and 4-hydroxy-2-oxoglutarate (HOG). As with PH types 1 and 2, PH type 3 is characterized by calcium-oxalate deposition in the kidneys or kidney stone formation. Most individuals with PH3 have early onset disease with recurrent kidney stones and urinary tract infections as common symptoms. Kidney failure is not a characteristic of PH3. Of note, individuals with heterozygous variants in *HOGA1* can have variable and intermittent elevations of urine oxalate. As with PH2, treatment for PH3 is focused on lowering calcium oxalate with increased daily fluid intake and inhibitors of calcium oxalate crystallization.

Secondary hyperoxalurias are due to hyperabsorption of oxalate (enteric hyperoxaluria); total parenteral nutrition in premature infants; ingestion of oxalate, ascorbic acid, or ethylene glycol; or pyridoxine deficiency and may respond to appropriate therapy.

A diagnostic workup in an individual with hyperoxaluria demonstrates increased concentration of oxalate in urinary metabolite screening. If glycolate, glycerate, or HOG is present, a primary hyperoxaluria is indicated. Confirmatory testing includes molecular analysis for PH1, PH2, or PH3 (CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies; refer to the Hyperoxaluria Diagnostic Algorithm for specific Gene List IDs).

Reference Values

Glycolate
< or =17 years: < or =75 mg/g creatinine
> or =18 years: < or =50 mg/g creatinine

Glycerate
< or =31 days: < or =75 mg/g creatinine
32 days - 4 years: < or =125 mg/g creatinine
5 - 10 years: < or =55 mg/g creatinine
> or =11 years: < or =25 mg/g creatinine

Oxalate
< or =6 months: < or =400 mg/g creatinine

7 months - 1 year: < or =300 mg/g creatinine
2 - 6 years: < or =150 mg/g creatinine
7 - 10 years: < or =100 mg/g creatinine
> or =11 years: < or =75 mg/g creatinine

4-hydroxy-2-oxoglutarate (HOG)
< or =10 mg/g creatinine

Interpretation

Increased concentrations of oxalate and glycolate indicate type 1 primary hyperoxaluria.

Increased concentrations of oxalate and glycerate indicate type 2 primary hyperoxaluria.

Increased concentrations of oxalate and 4-hydroxy-2-oxoglutarate indicate type 3 primary hyperoxaluria.

Increased concentrations of oxalate with normal concentrations of glycolate, glycerate, and 4-hydroxy-2-oxoglutarate indicate secondary hyperoxaluria.

Cautions

Ascorbic acid (vitamin C) will falsely elevate oxalic acid results.

Clinical Reference

1. Bhasin B, Urekli HM, Atta MG. Primary and secondary hyperoxaluria: Understanding the enigma. World J Nephrol. 2015;4(2):235-244. doi:10.5527/wjn.v4.i2.235
2. Michael M, Harvey E, Milliner DS, et al. Diagnosis and management of primary hyperoxalurias: best practices. Pediatr Nephrol. 2024;39(11):3143-3155. doi:10.1007/s00467-024-06328-2
3. Milliner DS, Harris PC, Cogal AG, et al. Primary hyperoxaluria type 1. In: Adam MP, Mirzaa GM, Pagon RA, et al, eds. GeneReviews [Internet]. University of Washington, Seattle; 2002. Updated August 15, 2024. Accessed September 23, 2025. Available at: www.ncbi.nlm.nih.gov/books/NBK1283/
4. Rumsby G, Hulton SA. Primary hyperoxaluria type 2. In: Adam MP, Mirzaa GM, Pagon RA, et al, eds. GeneReviews [Internet]. University of Washington, Seattle; 2008. Updated December 21, 2017. Accessed September 23, 2025. Available at: www.ncbi.nlm.nih.gov/books/NBK2692/
5. Milliner DS, Harris PC, Sas DJ, Lieske JC. Primary hyperoxaluria type 3. In: Adam MP, Mirzaa GM, Pagon RA, et al, eds. GeneReviews [Internet]. University of Washington, Seattle; 2015. Updated February 9, 2023. Accessed September 23, 2025. Available at: www.ncbi.nlm.nih.gov/books/NBK316514/
6. Fraser AD: Importance of glycolic acid analysis in ethylene glycol poisoning. Clin Chem. 1998;44(8):1769

Performance

Method Description

Urine samples corresponding to 0.25 mg of creatinine (not to exceed 1 mL of urine) are oximated to stabilize one of the target analytes, 4-hydroxy-2-oxoglutaric. The urine is then acidified and extracted. After evaporation, the dry residue is silylated and analyzed by capillary gas chromatography mass spectrometry.(Unpublished Mayo method)

PDF Report
No

Day(s) Performed
Wednesday

Report Available
3 to 9 days

Specimen Retention Time
2 months

Performing Laboratory Location
Mayo Clinic Laboratories - Rochester Main Campus

Fees & Codes

Fees
- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact Customer Service.

Test Classification
This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information
82542

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
HYOX	Hyperoxaluria Panel, U	53710-0

Result ID	Test Result Name	Result LOINC® Value
50592	Glycolate	13751-3
50593	Glycerate	13749-7
50594	Oxalate	13483-3
29984	Reviewed By	18771-6
29982	Interpretation	59462-2
38049	4-hydroxy-2-oxoglutarate	13678-8