Overview

Useful For
Differentiating between disorders of peroxisomal biogenesis (eg, Zellweger syndrome) and disorders with loss of a single peroxisomal function
Detecting abnormal elevations of pipecolic acid in urine

Genetics Test Information
Pipecolic acid is not detected by conventional organic acid analysis of urine.
In the newborn period, pipecolic acid levels are more likely to be abnormal in urine than in plasma or serum. Abnormal levels of pipecolic acid should be interpreted together with the results of other biochemical markers of peroxisomal disorders, such as plasma C22-C26 very long-chain fatty acids, phytanic acid, pristanic acid, RBC plasmalogens, and bile acid intermediates.

Highlights
Measurement of pipecolic acid is a useful diagnostic tool for differentiating between peroxisomal biogenesis disorders (Zellweger spectrum disorders) and peroxisomal disorders caused by single enzyme deficiencies such as X-linked adrenoleukodystrophy (X-ALD).

Results must be interpreted together with the results of other biochemical markers for peroxisomal disorders.
Both urine and plasma are suitable specimens for the detection of pipecolic acid.

Testing Algorithm
See Epilepsy: Unexplained Refractory and/or Familial Testing Algorithm in Special Instruction.

Special Instructions
- Epilepsy: Unexplained Refractory and/or Familial Testing Algorithm

Method Name
GasChromatography-MassSpectrometry(GC-MS)

NY State Available
Yes

Specimen

Specimen Type
Urine

Necessary Information
Patient’s age is required.

Specimen Required
Supplies: Plastic, 10-mL urine tube (T068)

Container/Tube: Plastic, 10-mL urine tube (T068)
Test Definition: PIPU
Pipecolic Acid, U

Specimen Volume: 5 mL

Collection Instructions:
1. Collect a random urine specimen.
2. No preservative.

Forms
If not ordering electronically, complete, print, and send an Inborn Errors of Metabolism Test Request (T798) with the specimen.

Specimen Minimum Volume
2 mL

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

Specimen Stability Information

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<th>Temperature</th>
<th>Time</th>
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<tr>
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<tr>
<td></td>
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Clinical and Interpretive

Clinical Information
Pipecolic acid (PA) is an intermediate of lysine metabolism and is oxidized in the peroxisomes by the enzyme L-pipecolate oxidase. In peroxisome biogenesis disorders (e.g., Zellweger syndrome), the activity of this enzyme is lost, resulting in an increase in pipecolic acid levels. In contrast, in peroxisomal disorders involving single enzyme deficiencies such as D-bifunctional protein deficiency, PA is not elevated; therefore PA analysis is useful for differentiating between these 2 groups of disorders.

Increased pipecolic acid levels may also be seen in alpha-aminoadipic semialdehyde dehydrogenase deficiency (pyridoxine dependent epilepsy), hyperlysinemia types 1 and 2, and defects in proline metabolism.

Theoretically, a defect in L-pipecolate oxidase can exist and several cases of hyperpipecolic acidemia have been reported, but a specific enzyme deficiency has not been described in any of the patients.

Reference Values
< or =31 days: < or =223.8 nmol/mg creatinine
32 days-5 months: < or =123.1 nmol/mg creatinine
6 months-11 months: < or =45.0 nmol/mg creatinine
> or =1 year: < or =5.7 nmol/mg creatinine
Interpretation

Elevated pipecolic acid levels are seen in disorders of peroxisomal biogenesis; normal levels are seen in disorders with loss of a single peroxisomal function.

Abnormal levels of pipecolic acid should be interpreted together with the results of other biochemical markers of peroxisomal disorders, such as plasma C22-C26 very long-chain fatty acids, phytanic acid, pristanic acid (POX / Fatty Acid Profile, Peroxisomal [C22-C26], Serum); RBC plasmalogens; and bile acid intermediates.

Cautions

Newborns with disorders of peroxisomal biogenesis often have normal levels of pipecolic acid that increase with age.

Abnormal results may reflect either prematurity or nongenetic liver and/or renal disease.

Pipecolic acid is not detected by conventional organic acid analysis (OAU / Organic Acids Screen, Urine).

Vigabatrin interferes with pipecolic acid determination.

Methylmalonic acid interferes with pipecolic acid determination.

Clinical Reference


Performance

Method Description


PDF Report

No

Day(s) and Time(s) Test Performed

Thursday; 8 a.m.

Analytic Time

Document generated October 29, 2019 at 9:59pm CDT
2 days

**Maximum Laboratory Time**

31 days

**Specimen Retention Time**

1 month

**Performing Laboratory Location**

Rochester

### Fees and 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 Regional Manager. 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. This test has not been cleared or approved by the U.S. Food and Drug Administration.

#### CPT Code Information

82542

#### LOINC® Information

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