

Organic Acid Test Information

Organic acidurias is a term to describe disorders of organic acid metabolism in humans, encompassing a variety of inborn errors of metabolism (IEM). Organic acidurias are often inherited metabolic diseases, however sometimes they can be acquired via nutritional, iatrogenic, and environmental sources. They often arise from a block in certain metabolic pathways and/or activation of alternate metabolic pathways by a specific enzyme deficiency, and can be characterized by accumulation of unusual organic acid intermediary products which are then excreted in urine.

Urinary organic acid screening is a powerful clinical tool in both diagnostic and long-term treatment of IEM due to organic acidurias. For example, it is crucial for early detection of diagnosis of acutely ill newborn and infants for suspected metabolic diseases, whom often have life-threatening symptoms. GC-MS is the gold standard for urinary organic acid analysis and has been so for several decades. The method can detect more than 250 organic acids in urine.

Our lab offers a sensitive and accurate GC-MS assay for urinary organic acids based on an ethyl acetate extraction/trimethylsilyl (TMS) derivatization method. The results are reported to and interpreted by Dr. Nancy Leslie, Dr. Carlos Prada, and Dr. Kathryn Nicole Weaver in the Clinical Biochemical Genetics Laboratory at Cincinnati Children's Hospital Medical Center (CCHMC). For more information, call the lab at 513-636-4203.

Sample Type:

Urine

Volume:

5.0 mL

2.5 mL (minimum)

Specimen Preparation:

Random urine collection. Freeze ASAP.

Stability:

Refrigerated: Up to 3 days

Frozen: 1 month

Methodology:

Gas chromatography-mass spectrometry (GC-MS)

Reporting Units:

None, qualitative

Reference Interval:

None, qualitative interpretation. Clinical information is needed for appropriate interpretation. Additional required information includes age, gender, diet, drug therapy and family history.

Shipping Conditions:

Frozen (dry ice), next day.

Testing Schedule:

Mon, Thu, 1st Shift (for testing outside this schedule, please call 513-636-4203). **Turnaround time:** 7 – 10 days.

CPT Codes:

83919

Contact Information:

Clinical Mass Spectrometry

Tel: 513-636-4203

Fax: 513-803-5014

Email: pathology@cchmc.org

Website: www.cincinnatichildrens.org/mass-spec

Shipping Address:

Clinical Mass Spectrometry Facility, MLC 7019
Department of Pathology and Laboratory Medicine
Cincinnati Children's Hospital Medical Center
240 Albert Sabin Way
Cincinnati, Ohio 45229-3039

References:

1. Chalmers RA. and Lawson AM, eds. Organic acids in man. London: Chapman & Hall, 1982.
2. Dagleish CE. et al. A gas-liquid-chromatographic procedure for separating a wide range of metabolites occurring in urine or tissue extracts. *Biochem. J.* 1966 (101) 792-810.
3. Kumps A. et al. Metabolic, nutritional, iatrogenic, and artefactual sources of urinary organic acids: a comprehensive table. *Clin. Chem.* 2002 (48) 708-717.