

## ORGANIC ACIDS (qualitative)

### Relevant disorders

Urea cycle disorders, fatty acid oxidation disorders, organic acidaemias, amino acidopathies, pyrimidine disorders,

### Related Metabolic Tests

Plasma amino acids, urine amino acids, quantitative orotic acid, quantitative methylmalonic acid, quantitative VMA/HVA, plasma acylcarnitines, dried blood spot acylcarnitines

### Indication for Test

Qualitative analysis of Organic Acids in urine can be used to diagnose, or aid in the diagnosis, of a variety of inherited metabolic disorders, including the classical organic acidaemias, fatty acid oxidation disorders, pyrimidine disorders, urea cycle defects and some amino acidopathies. Interpretation is based on the presence of certain key metabolites combined with pattern recognition. In some disorders organic acid analysis may be essentially diagnostic, e.g. propionic acidaemia or  $\beta$ -ketothiolase deficiency, but in other cases the presence of a particular metabolite may point towards a group of disorders without being specific e.g. the presence of orotic acid in urea cycle disorders.

### Methodology

Organic acids are extracted from acidified, salt saturated urine. The extracts are evaporated to dryness under nitrogen and trimethylsilyl (TMS) derivatives formed using  $N_2O$ -bis(trimethylsilyl)trifluoroacetamide (BSTFA) and pyridine. TMS derivatives are identified using gas chromatography mass spectrometry (GCMS) electron impact.

### Sample requirements

10mL aliquot of a random urine or a 24 hour urine in a plain bottle. DO NOT USE borate. Smaller samples are accepted if necessary.

### Turn Around Time

5 – 14 days (but if requested urgently can be performed more quickly).

## Transport information/ Contact Details

Send by first class post.

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## References

- Diagnosis of Organic Acidaemias by Gas Chromatography Mass Spectrometry. Goodman and Markey, Alan R Liss, Inc., New York, 1981.
- Organic Acids in Man: Analytical Chemistry, Biochemistry and Diagnosis of the Organic Acidurias. Chalmers and Lawson, Chapman and Hall, 1982.