

ETHYLMALONATE

Relevant disorders

Ethylmalonic acidaemia, GA2, SCADD

Related Metabolic Tests

Urinary organic acids
Acylcarnitines

Indication for Test

Quantitative measurement of EMA excretion is used:

- 1 To determine the significance of an apparently elevated excretion on qualitative organic acid analysis, this must be interpreted in light of the clinical details.
- 2 To monitor patients with Ethylmalonic acidaemia, SCADD and Glutaric acidaemia Type II.

Methodology

Stable isotope dilution GC-MS.

Sample requirements

5 mL random urine (no preservative).

Transport information/ Contact Details

Send by first class post. Normal packaging.

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Turn Around Time

2 weeks for urine analysis

Reference Ranges

< 15 $\mu\text{mol}/\text{mmol}$ creatinine

Reference range established internally from patient data

(N.B: reference range only valid with data produced by the Department of Clinical Chemistry, Sheffield Children's NHS Foundation Trust).

Samples from patients with confirmed diagnoses of EMA and GA2 have also been analysed.

| Disorder | EMA: creatinine ratio ($\mu\text{mol}/\text{mmol}$ creatinine) | |
|--|--|-------|
| Ethylmalonic encephalopathy (1 patient, 2 separate samples) | 188.9 | 166.3 |
| GA2 (1 patient, 2 separate samples) | 198.3 | 142.2 |