

## ISOVALERYLGLYCINE

### Relevant disorders

Isovaleric acidaemia

### Related Metabolic Tests

Urine organic acids  
Acylcarnitines

### Indication for Test

Following a presumptive positive newborn screening result for Isovaleric Acidaemia (IVA).

### Methodology

Tandem mass spectrometry

### Sample requirements

2ml random urine, no preservative

### Transport information/ Contact Details

Send by first class post.

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### Turn Around Time

ASAP. Metabolic laboratory MUST be contacted (see contact information below).

## Reference Ranges

Unaffected = < 2.6  $\mu\text{mol}/\text{mmol}$  creatinine

## References

Ensenauer R et al (2004) A Common Mutation Is Associated with a Mild, Potentially Asymptomatic Phenotype in Patients with Isovaleric Acidemia Diagnosed by Newborn Screening. *American Journal of Human Genetics* 75: 1136 – 1142