SULPHOCYSTEINE - URINE (quantitative)

Relevant disorders

Isolated Sulphite Oxidase deficiency
Molybdenum cofactor deficiency (combined sulphite oxidase and xanthine oxidase deficiency)

Related Metabolic Tests

Amino acids – urine (quantitative)
Amino acids – plasma (quantitative)
Urate – plasma and urine
Xanthine and hypoxanthine – urine
Thiosulphate - urine
Sulphite - urine

Indication for Test

Sulphocysteine (SCYS) is a pathognomic metabolite found in large quantities in the urine of patients with the inherited metabolic disorders isolated sulphite oxidase deficiency and molybdenum cofactor deficiency (combined sulphite oxidase and xanthine oxidase deficiency due to lack of the required molybdenum cofactor) but is found in only trace amounts in individuals not affected by these disorders. These patients present with severe infantile epileptic encephalopathy and progressive psychomotor retardation. Elevated urinary levels of sulphite, thiosulphate and sulphocysteine are hallmarks in the diagnosis of sulphite oxidase deficiency and molybdenum cofactor deficiency. However, sulphite testing has to be performed on very fresh urine (i.e. at the bedside) and the assay for thiosulphate has many known interferences. The rapid progression of the disease symptoms in non-treated patients means an early diagnosis and monitoring of treatment efficacy is crucial in aiding a good treatment outcome in patients with molybdenum cofactor deficiency type A.

Isolated sulphite oxidase deficiency is less common that molybdenum cofactor deficiency and can be excluded if the products of other molybdenum cofactor dependent enzymes (such as uric acid) are reduced in urine and plasma and urinary xanthine and hypoxanthine are elevated.

Methodology

Tandem mass spectrometry – stable isotope dilution method

Sample requirements

5ml urine in a plain universal container

Turn Around Time

1 week (but if requested urgently can be performed more quickly).

Transport information/Contact details

Send all samples by first class post or courier if urgent to:

Department of Clinical Chemistry
Sheffield Children's NHS Foundation Trust
Western Bank, Sheffield
S10 2TH, UK

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References

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