DEPARTMENT OF CLINICAL CHEMISTRY
SHEFFIELD CHILDREN'S NHS FOUNDATION TRUST

AMINO ACIDS - CSF (quantitative)

Relevant disorders

Non ketotic hyperglycinaemia (NKH, Glycine encephalopathy)
Serine deficiency disorders

Related Metabolic Tests

Plasma amino acids
Organic acids (if increased glycine measured in plasma, as part of differential diagnosis)

Indication for Test

Serine, glycine, threonine and alanine can be measured in CSF.

NKH is usually classified into 2 main clinical types: neonatal and late-onset. The neonatal onset is the most common. Symptoms include muscular hypotonia, seizures, apnoeic attacks, lethargy and coma.
Increased glycine concentrations in CSF and plasma are biochemical features of the disorder.

Serine deficiency disorders are very rare. Includes 3-phosphoglycerate dehydrogenase deficiency, 3-phosphoserine aminotransferase deficiency, 3-phosphoserine phosphatase deficiency. Patients with 3-phosphoglycerate dehydrogenase deficiency are affected with confgenital microcephaly, psychomotor retardation and intractable seizures.

Methodology

Quantitative amino acid analysis is performed on a Biochrom amino acid analyser which uses a cation exchange resin and Ninhydrin detection.

Sample requirements

Plain CSF sample (fluoride acceptable), 0.2ml. **Sample must not be blood stained**.
Paired lithium heparin plasma sample also required, 1ml Lithium heparin plasma (Fluoride and EDTA acceptable).

Please note: if a serine deficiency is suspected the plasma sample should be taken while fasting. Plasma serine levels can be normal after feeding.

Turn Around Time

5 – 14 days (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

Joanne Croft (Clinical Scientist)
0114 2717307

Reference Ranges

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

Glycine

CSF:plasma ratio:	0.01 – 0.04 normal
	0.09 – 0.25 neonatal NKH
	0.06 – 0.1 late onset NKH

Please note: the diagnosis of NKH can only be confirmed by enzyme assay in liver tissue.

Serine

(taken from Fernandes, Saudubray, van den Berghe, Walter. Inborn Metabolic Diseases, Diagnosis and Treatment, 4th Edition. Published by Springer, 2006.)

Control range: 35 – 80µM
Affected: 6 – 8 µM

References

- Fernandes, Saudubray, van den Berghe, Walter. Inborn Metabolic Diseases, Diagnosis and Treatment, 4th Edition. Published by Springer, 2006.