DEPARTMENT OF CLINICAL CHEMISTRY
SHEFFIELD CHILDREN'S NHS FOUNDATION TRUST

AMINO ACIDS - HAIR (quantitative)

Relevant disorder

Trichothiodystrophy Syndrome

Indication for Test

Trichothiodystrophy (TTD) is a term relating to sulphur-deficient brittle hair in patients presenting with mental and physical retardation as well as ichthyotic skin. Reduced cystine in the matrix proteins of hair and nails results in fragility and morphological abnormalities. Approximately half of TTD patients display photosensitivity due to a nucleotide excision repair defect, although no cases of skin cancer have been reported.
Diagnosis of TTD can be made by analysing the amino acid content of hair.

Methodology

A sample of hair is hydrolysed with hydrochloric acid in an oxygen-free environment for 30 hours at high temperature. The resultant solution is purified and analysed by ion exchange liquid chromatography using the ninhydrin reaction and UV absorption for detection and quantitation.

To aid the biochemical diagnosis, the abnormal morphology of TTD hair can exhibit birefringence (striped 'tiger tail' pattern) and trichorrhexis nodosa (distorted shaft with 'nodes').

Sample requirements

Hair sample from area of brittle hair – as much as possible (50mg at least, equivalent to a small 'lock' of hair).
Stored at room temperature prior to analysis.

Transport information/Contact details

Send all samples by first class post to:

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

Joanne Croft (Clinical Scientist)
0114 2717307

Turn Around Time

2 months

Reference Ranges

Based on 2 standard deviations about the mean of 3 control adult analyses.

Amino Acid	Percentage of total amino acid content (%)		
Aspartic acid	5.5 – 6.7		HIGH for TTD
Threonine	7.7 – 8.9		LOW for TTD
Serine	11.3 – 13.5		LOW for TTD
Glutamic acid	12.4 – 15.0		(not sig.)
Proline	6.4 – 10.6		LOW for TTD
Glycine	6.1 – 6.7		(not sig.)
Alanine	4.8 – 5.6		HIGH for TTD
Valine	5.1 – 6.5		(not sig.)
CYSTINE	7.4 – 10.6	(very significant marker)	LOW for TTD
Methionine	0.3 – 0.7		(not sig.)
Isoleucine	2.1 – 3.3		(not sig.)
LEUCINE	6.5 – 8.1	(very significant marker)	HIGH for TTD
Tyrosine	1.7 – 2.5		(not sig.)
Phenylalanine	1.6 – 2.2		HIGH for TTD
Histidine	0.5 – 1.3		(not sig.)
Lysine	1.8 – 3.2		HIGH for TTD
Arginine	6.6 – 7.8		(not sig.)

References

- Price VH et al Trichothiodystrophy. (1980) Sulfur-deficient hair as a marker for a neuroectodermal symptom complex. Arch. Derm. 116: 1375-1384

- Itin PH & Pittlekow MR (1990) Trichothiodystrophy: review of sulfur-deficient brittle hair syndromes and association with the ectodermal dysplasias. Am.Acad.Derm 22: 705-717.

- Lehman AR et al (1994) Nomenclature of human DNA repair genes. Mutat.Res.315: 41-42.

- Pollitt RJ et al (1968) Sibs with mental and physical retardation and trichorrhexis nodosa with abnormal amino acid composition of the hair. Arch. Dis. Child Vol 43 No 228 211-216