

Case Report

Dr Mick Henderson

Biochemical Genetics

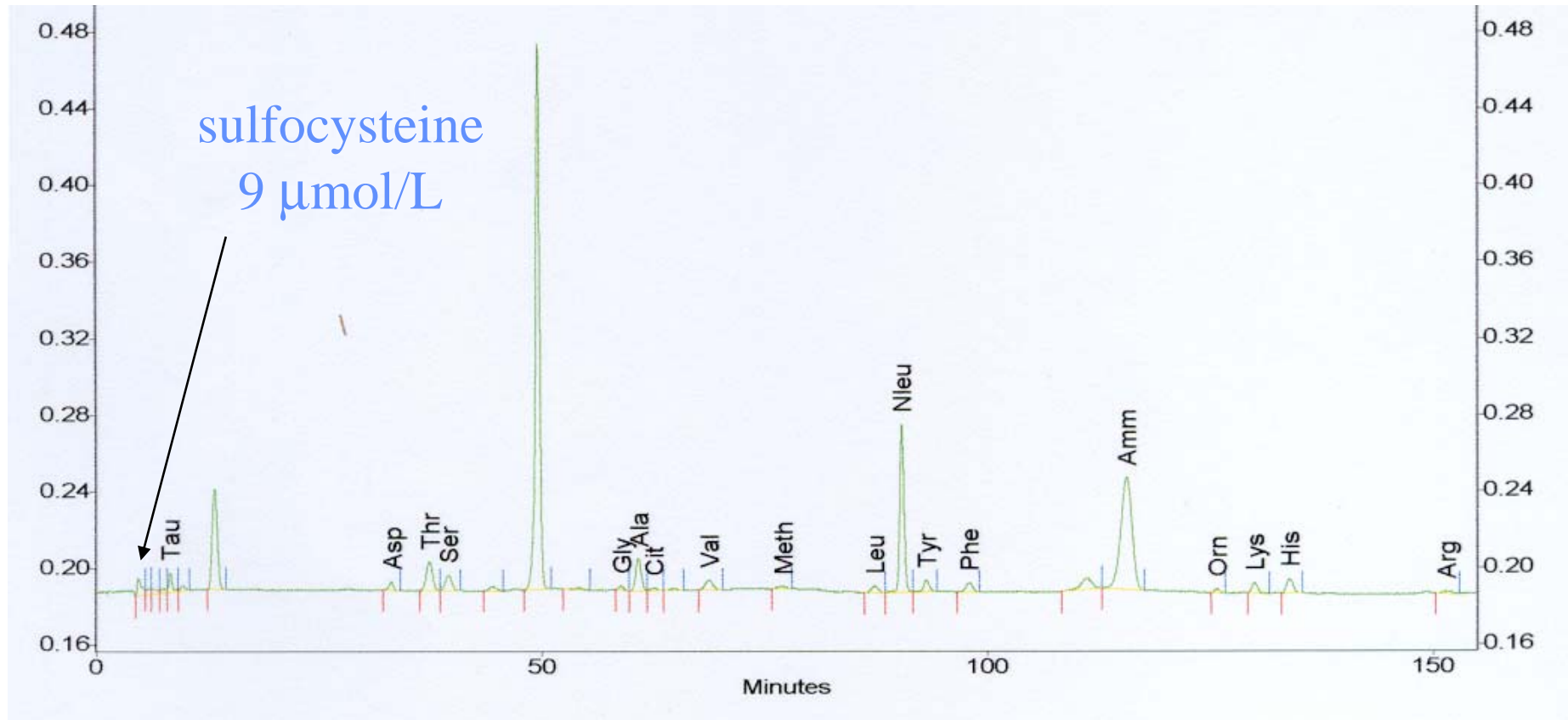
Department of Clinical Biochemistry

Leeds Teaching Hospitals

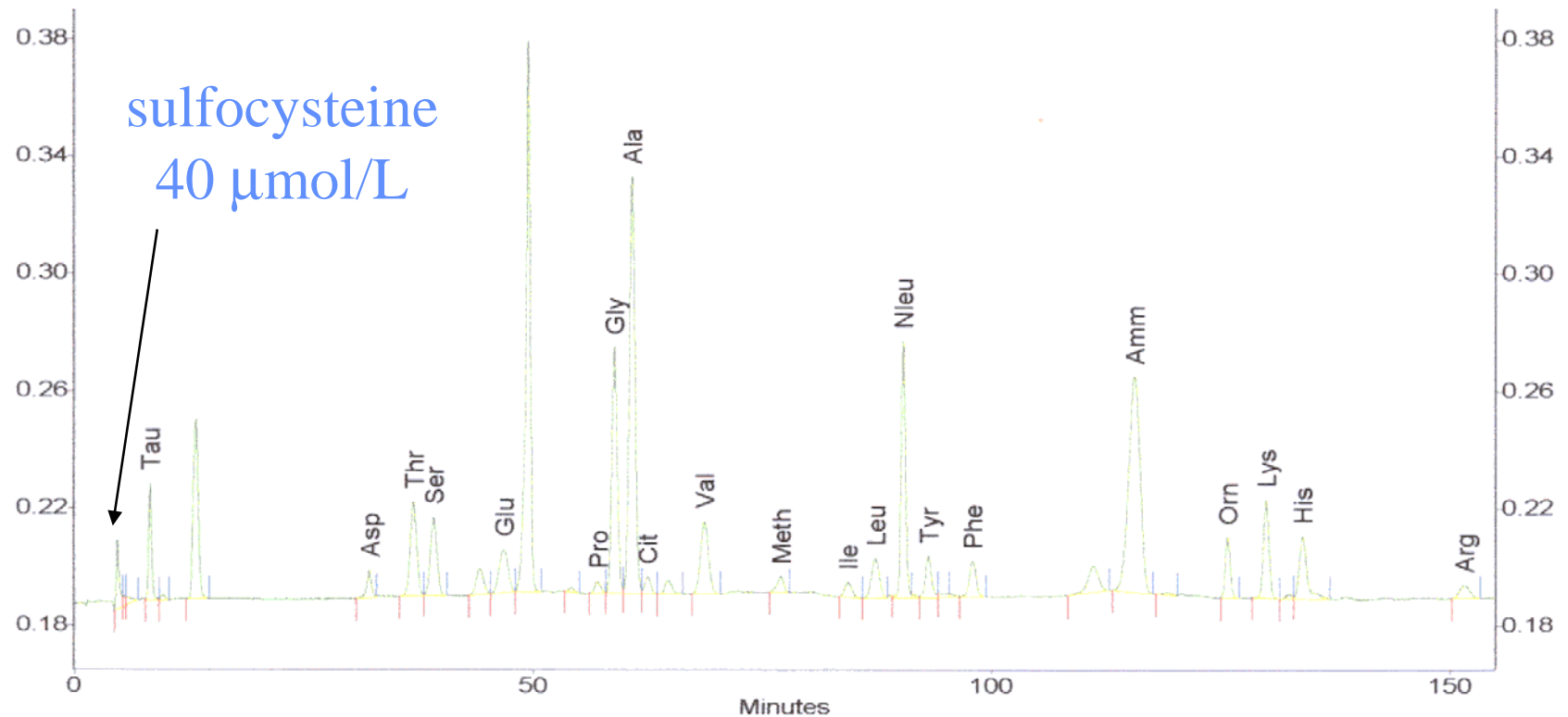
Female baby

- unrelated parents
 - term baby, no recorded neonatal problems
 - severe persistent fitting from day 2
 - died at 3 weeks
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- early apnoea, lactate 8 mmol/L
 - no evidence of hyperammonaemia, hypoglycaemia
 - urine organic acids & blood acyl carnitines: NAD

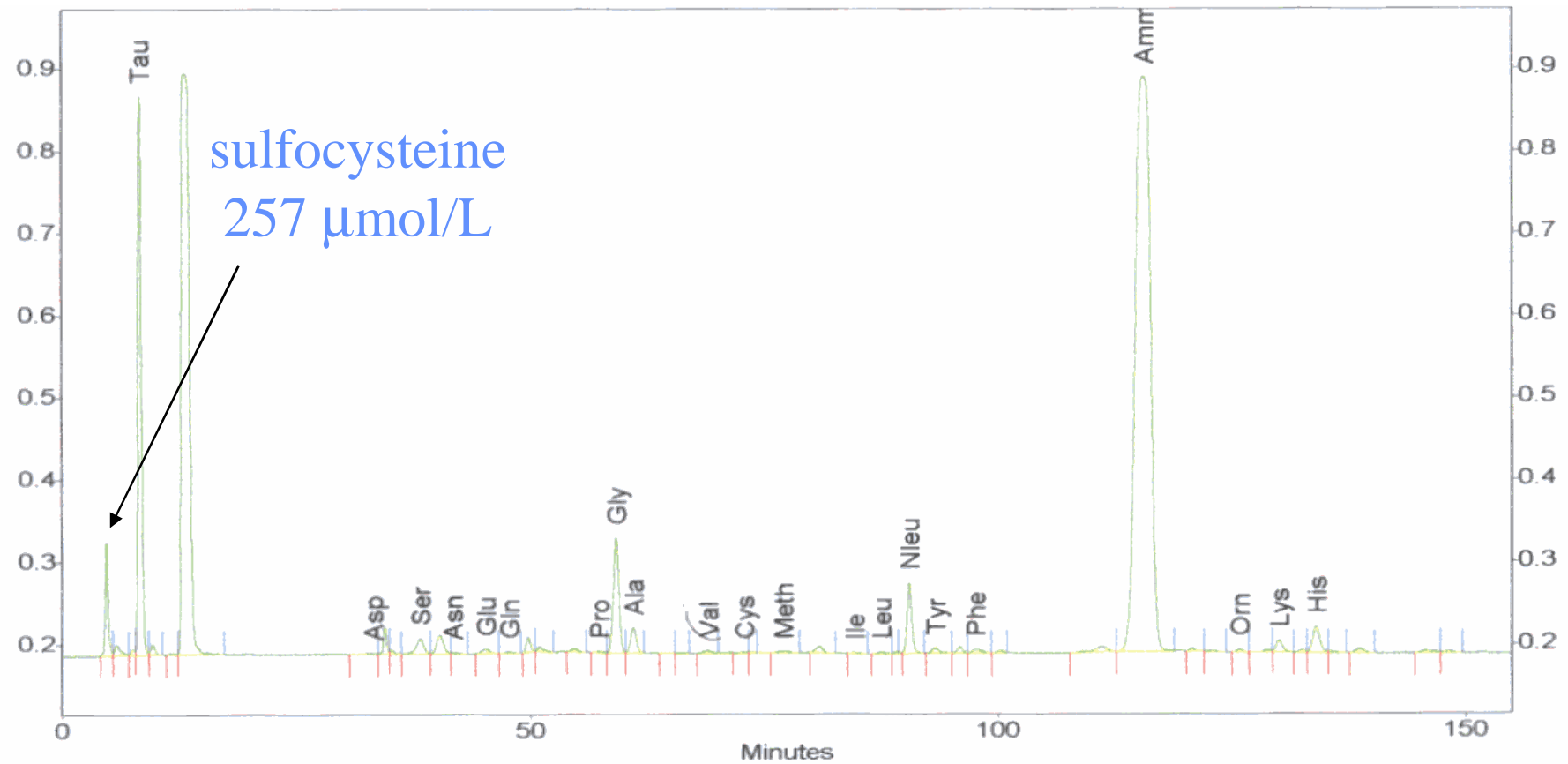
CSF amino acids



Plasma amino acids



Urine amino acids



Purine Metabolism

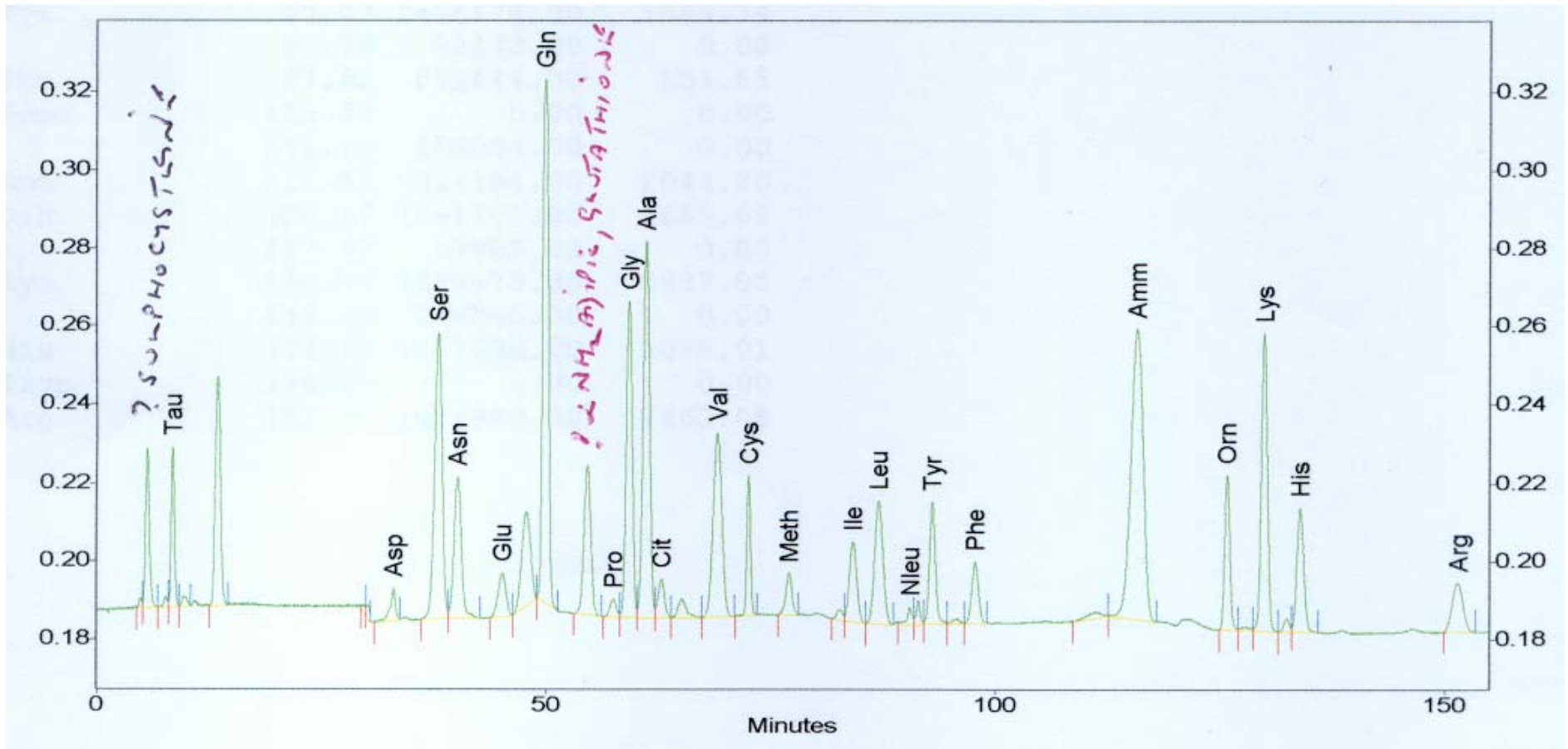
- Plasma urate: 0.18 (*ref 0.14-0.26*)
- Urine urate:creatinine: 1.18 & 0.71 (*ref 0.43-1.52*)
- Report from Purine Lab at Guy's:

no evidence of molybdenum cofactor deficiency

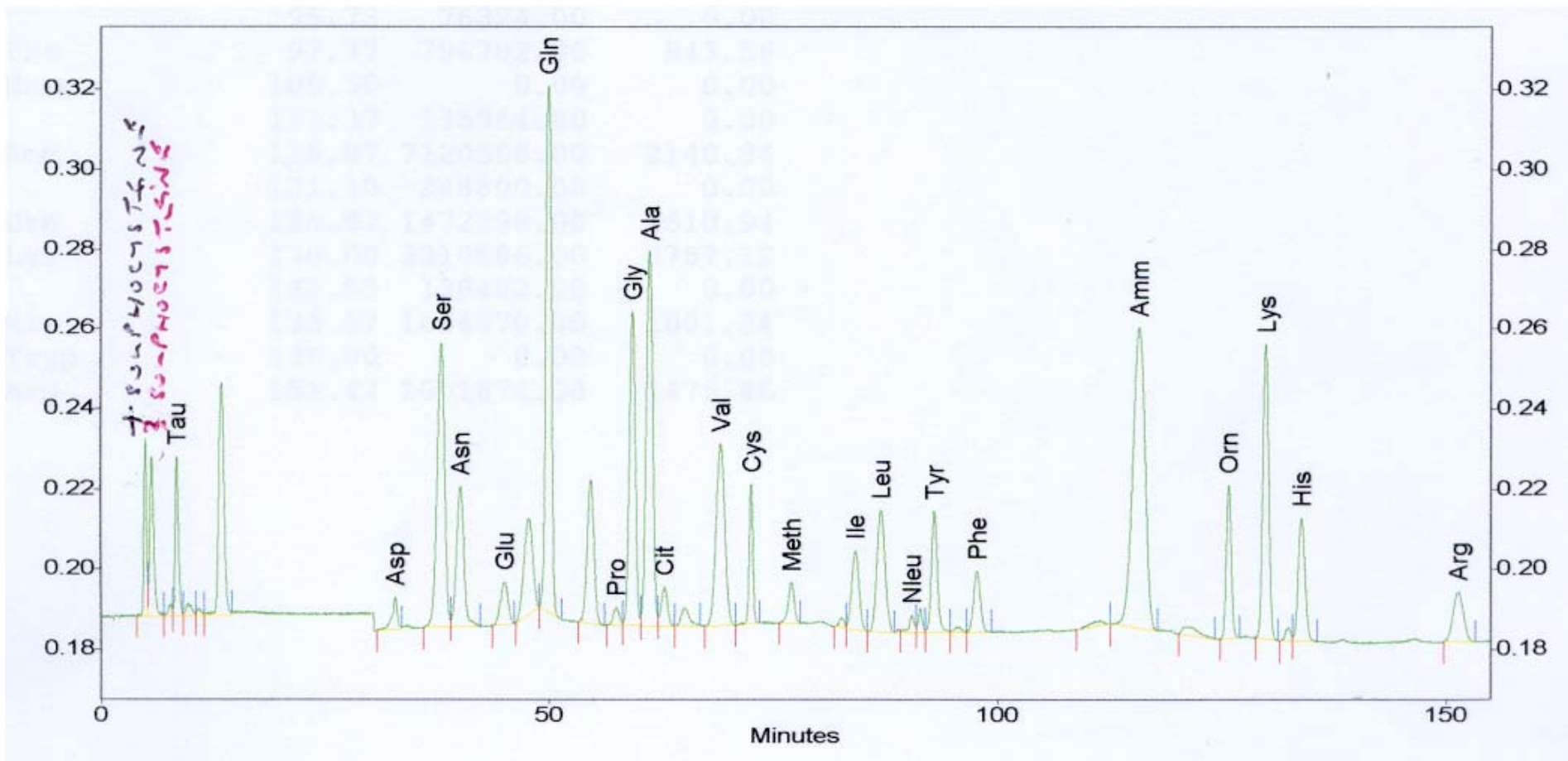
Results summary

Date	Urine						Plasma			
	sulfocys	taurine	cystine	glycine	sulphite		sulfocys	taurine	cystine	glycine
<i>ref value</i>	<i>ND</i>	<i><1051</i>	<i><37</i>	<i><938</i>	<i>neg</i>		<i>ND</i>	<i>92-392</i>	<i>21-73</i>	<i>220-527</i>
6.8.00	139	448	3	504	neg		40	76	ND	244
14.8.00							55	298	ND	449
15.8.00							46	308	ND	412
17.8.00	356	1067	19	2070	pos		44	319	ND	438
22.8.00							60	112	ND	288
24.8.00	304	2087	6	557	neg		40	148	ND	256
25.8.00	367	2404	11	591	neg					

Plasma amino acids, referred sample



Plasma amino acids, referred sample spiked with sulphocysteine



570:440 ratios, scys 4.3, unknown 10.0

Biochemical Genetics LTHT

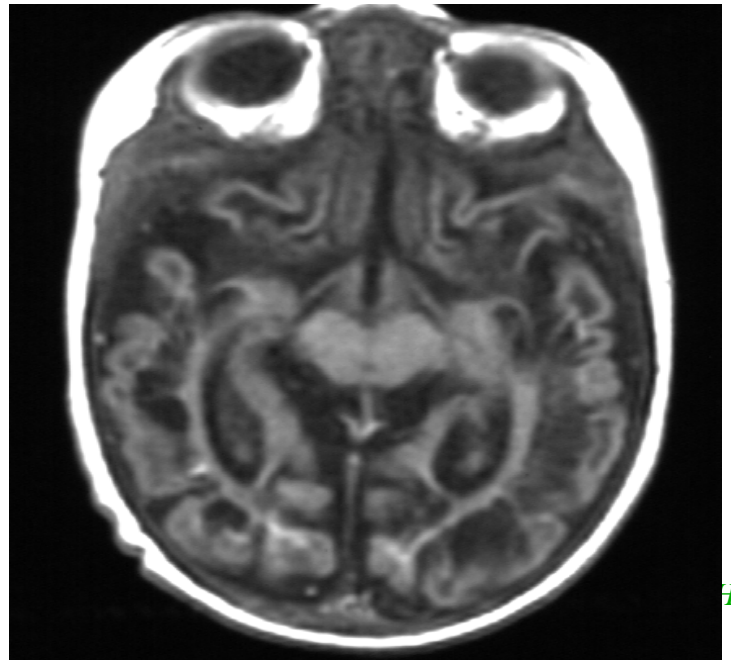
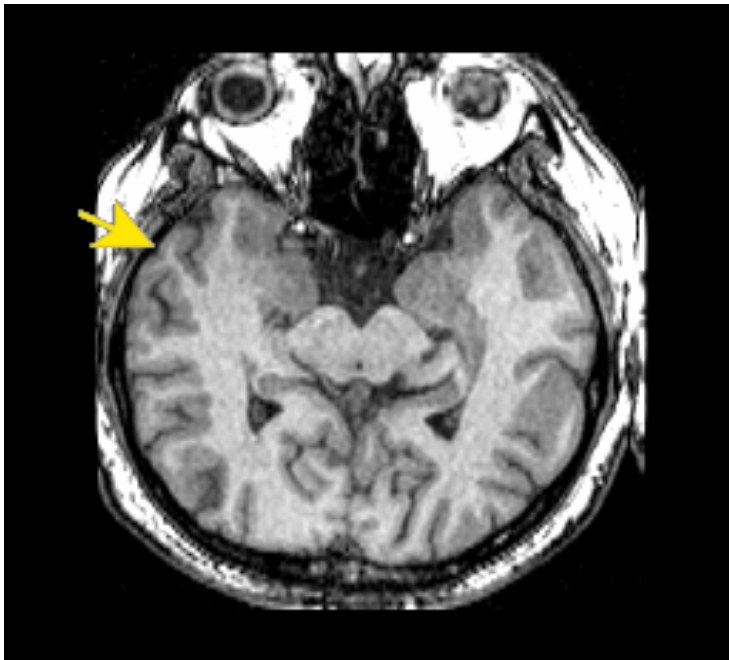
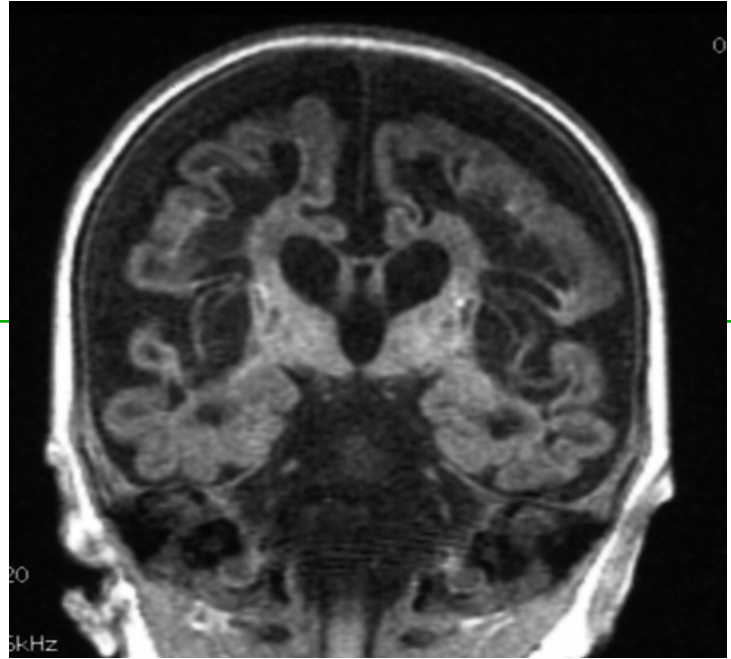
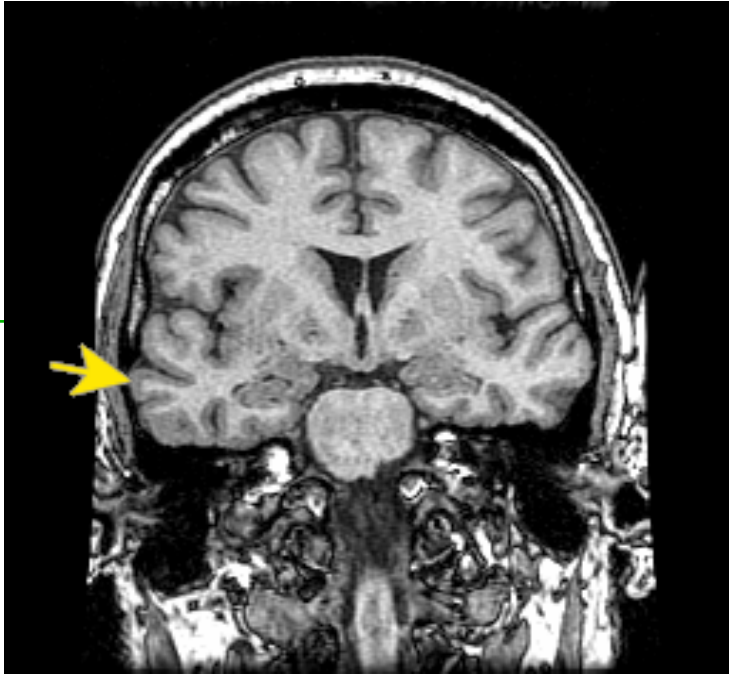
Molybdenum Cofactor deficiency

Case 2

- Baby of second cousin parents.
- Uncomplicated pregnancy.
- Ultrasound scan at 23 weeks gestation showing normal foetus and brain.
- Uncomplicated normal delivery at 39+ weeks.

Post-partum

- 12h post partum – abnormal movements, high pitched cry, lethargy, hypertonic, pyrexial, not feeding and fits by day 1.
- Admitted to SCBU



Further investigations

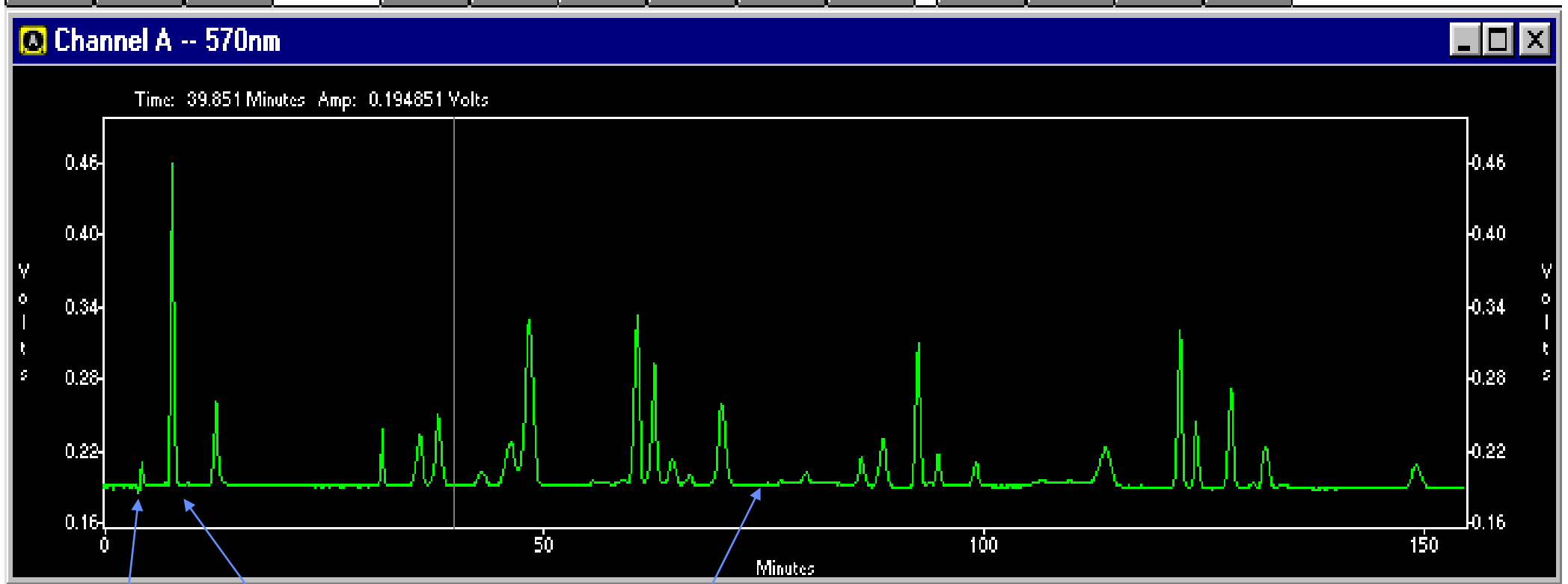
- Following scans, usual biochemistry and pathology work up investigations carried out.
- Baby's condition worsened and developed severe seizures.
- Started on Phenobarbitone with little benefit.

Urine Purines

Urate	0.000 mmol/L
Hypoxanthine	0.112 mmol/L
Xanthine	2.076 mmol/L
Urate/Creat ratio	0.00 (0.30-1.50)
Pseudouridine	0.696 mmol/L

- Results consistent with xanthine oxidase deficiency.

Plasma



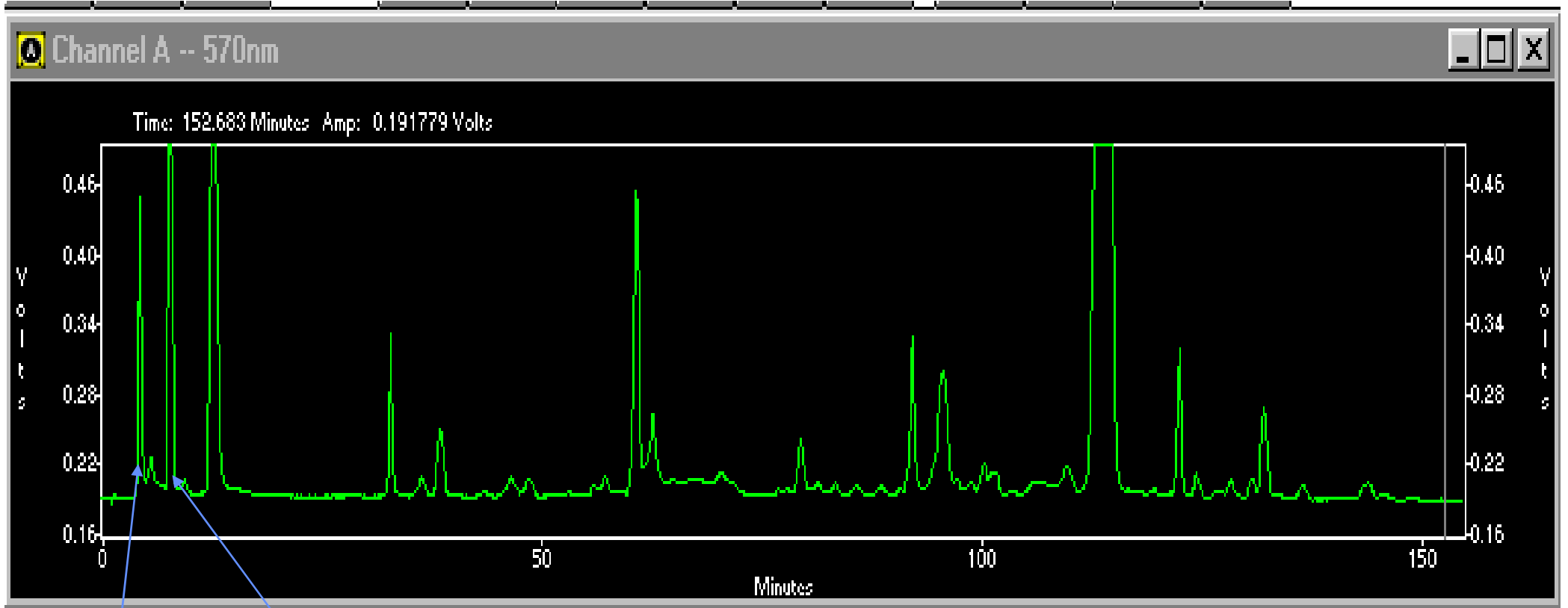
sulphocysteine

taurine

no cystine

◆ Total plasma homocysteine undetectable.

Urine

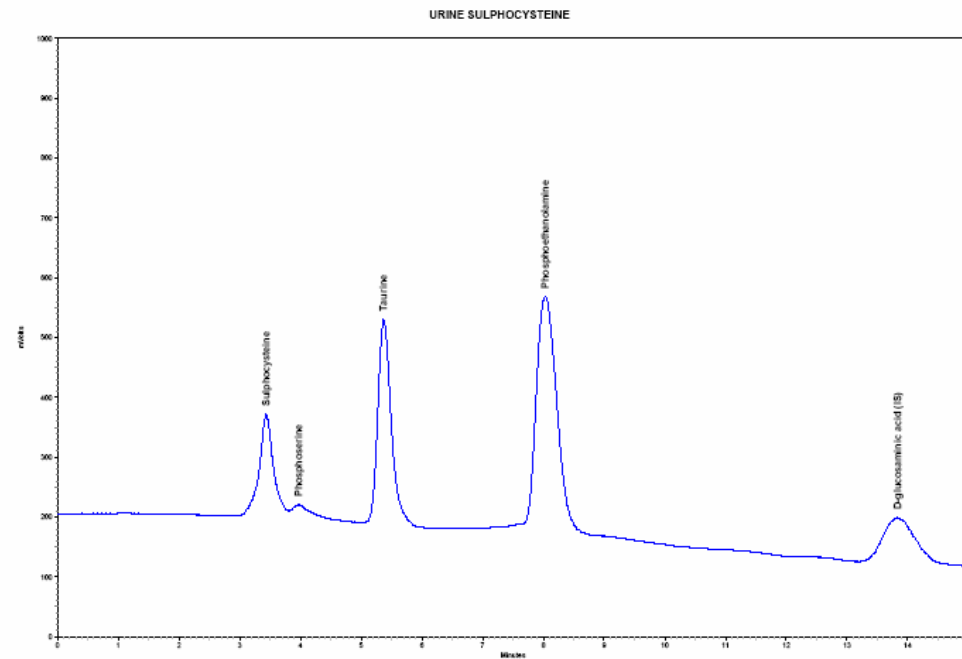


sulphocysteine

taurine

Urine; Case 3, 2005

New short programme for Biochrom

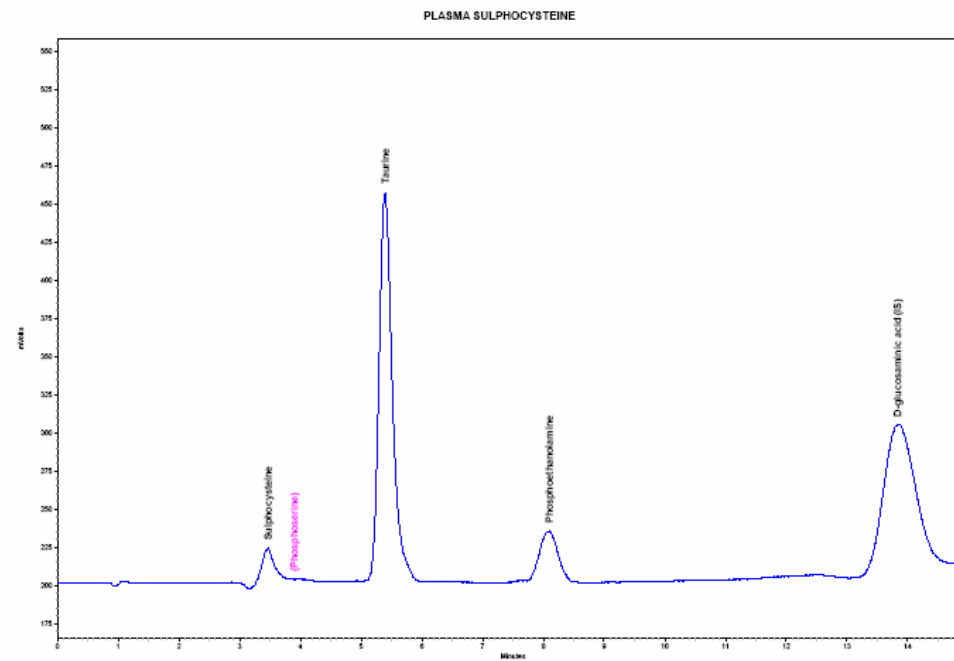


Ref: Applications note Bio30.4 - Rapid analysis of sulphocysteine

Data kindly provided by St James's Hospital, Leeds UK

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