

# Case Report

---

***Dr Mick Henderson***

*Biochemical Genetics*

*Department of Clinical Biochemistry*

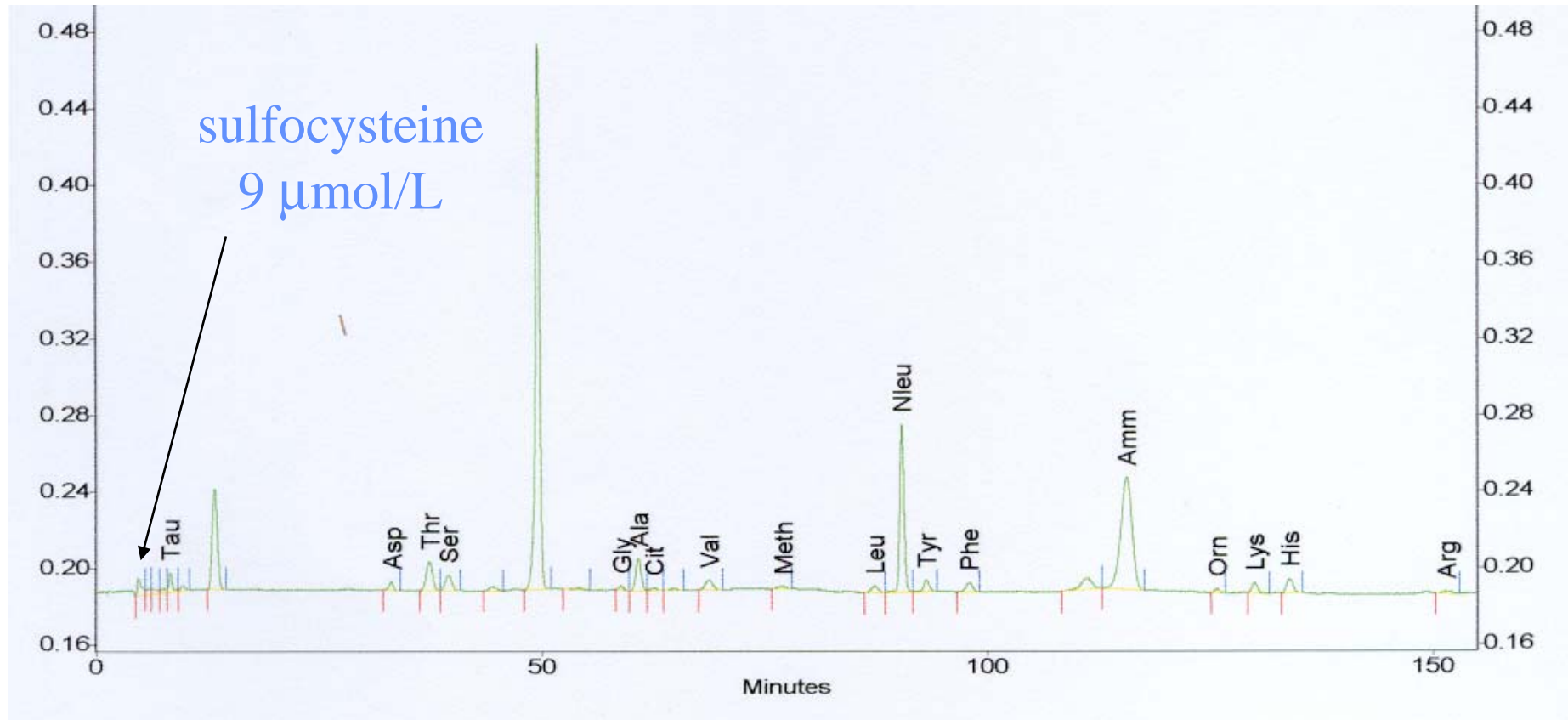
*Leeds Teaching Hospitals*

# Female baby

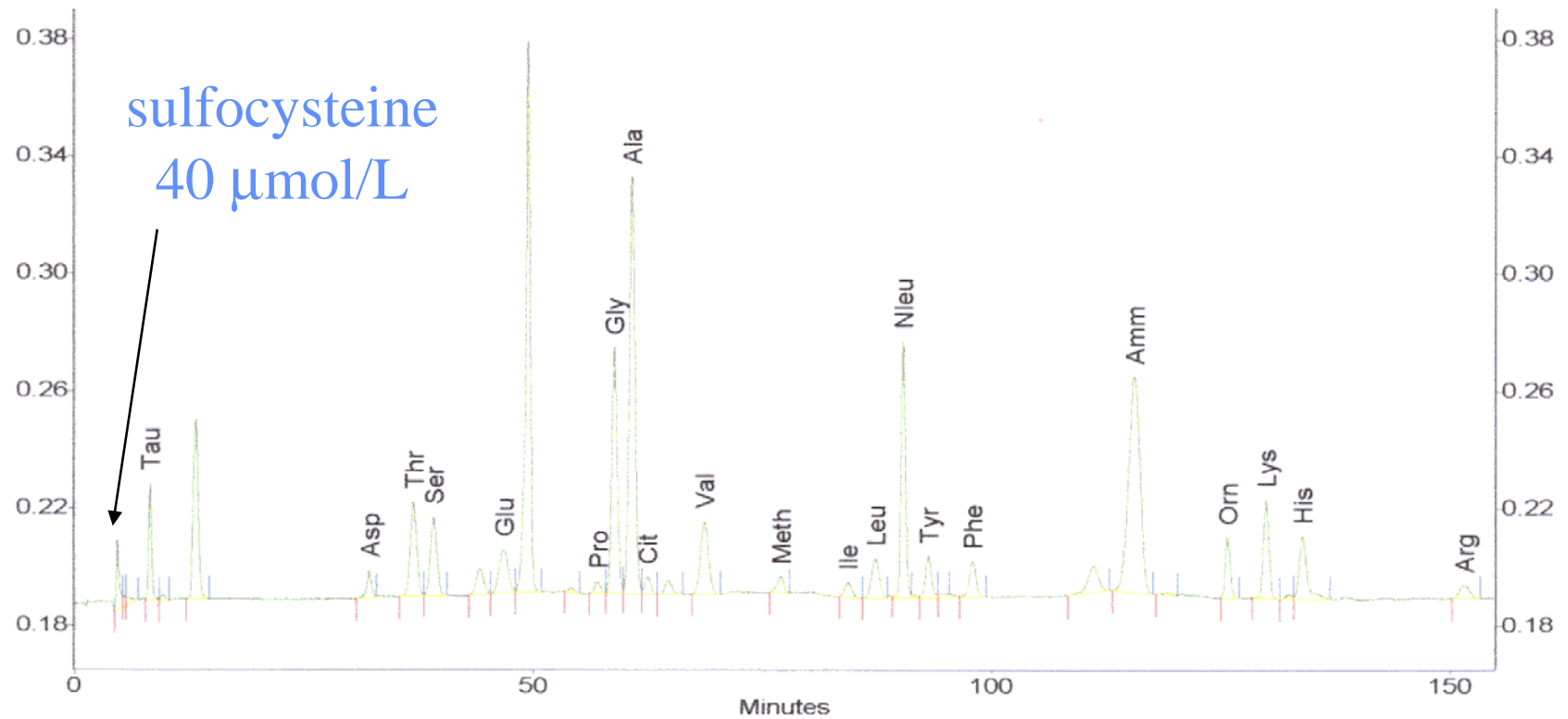
---

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

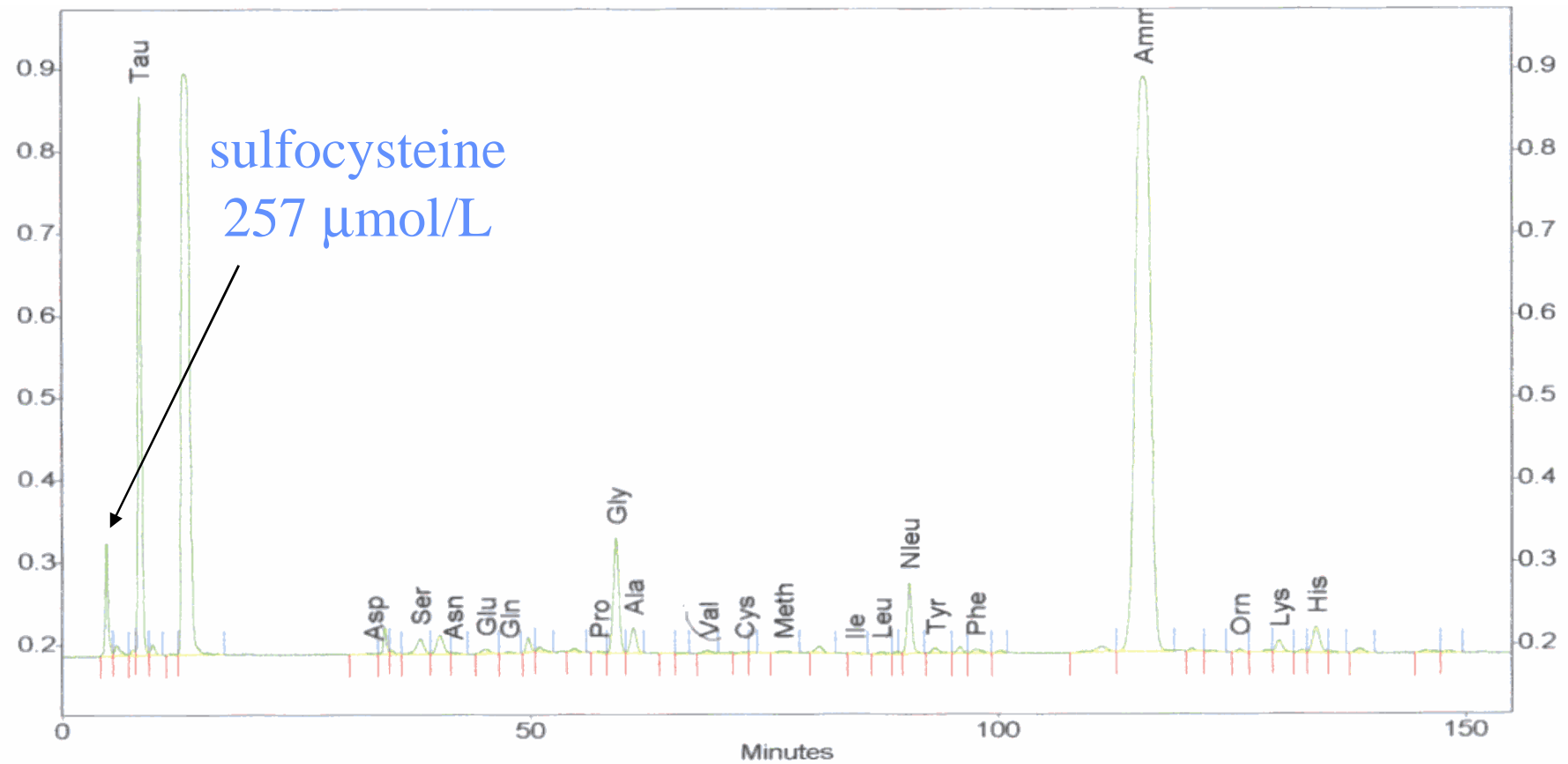
# CSF amino acids



# Plasma amino acids



# Urine amino acids



# Purine Metabolism

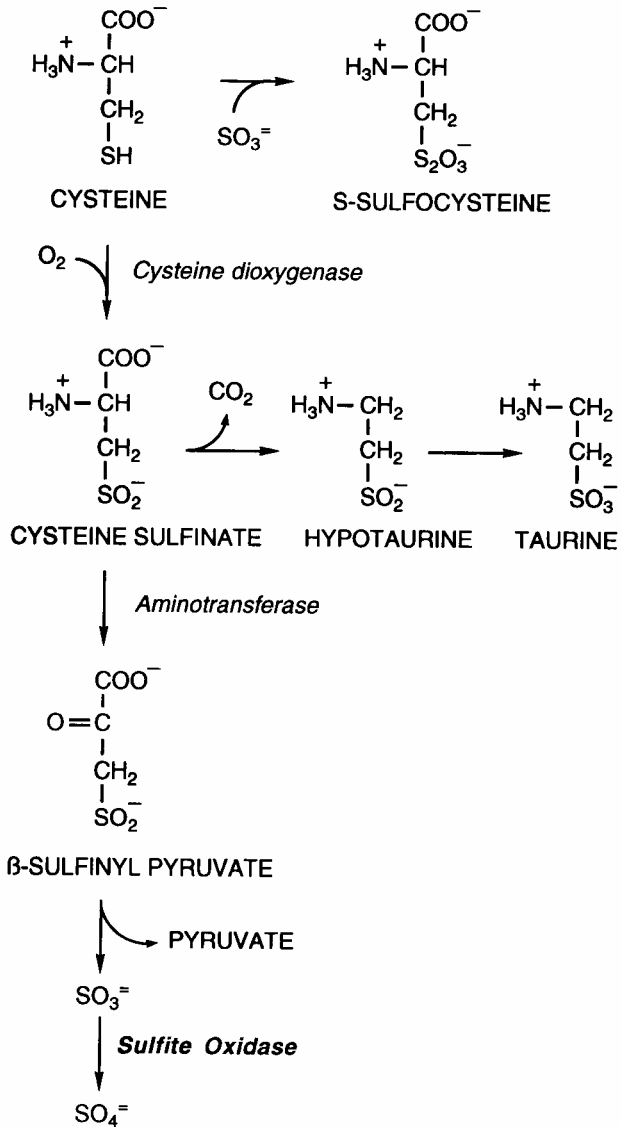
---

- 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:

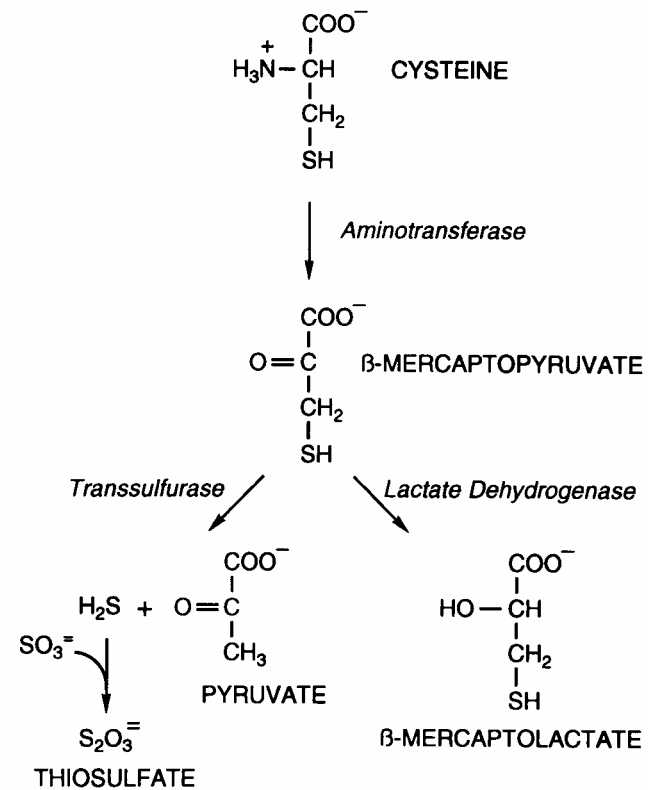
*no evidence of molybdenum cofactor deficiency*

# Results summary

Date	Urine						Plasma			
	sulfocys	taurine	cystine	glycine	sulphite		sulfocys	taurine	cystine	glycine
<i>ref value</i>	<i>ND</i>	<i>&lt;1051</i>	<i>&lt;37</i>	<i>&lt;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					



**FIG. 70-6** Major pathway of cysteine catabolism: direct oxidation. Formation of S-sulfocysteine.



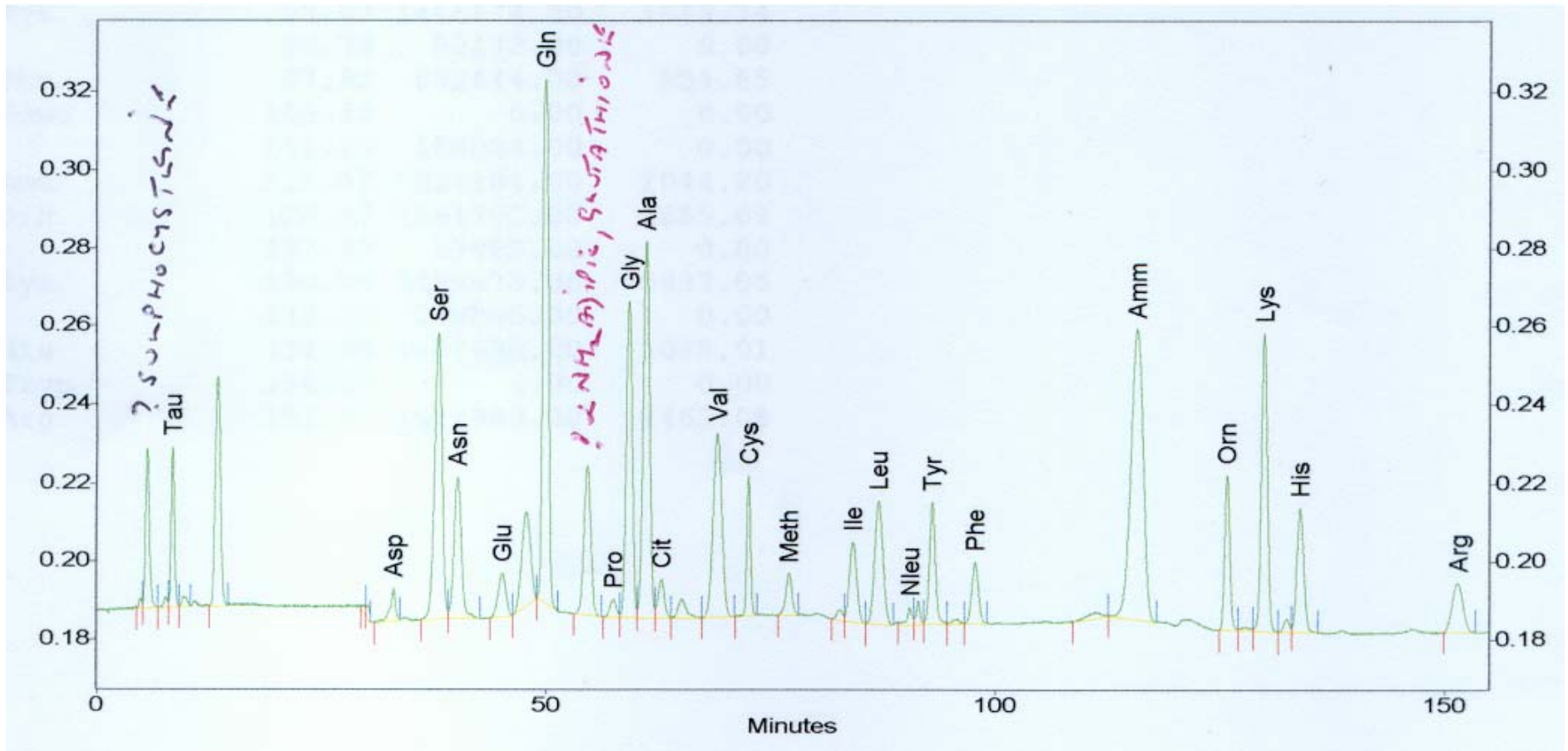
**FIG. 70-7** Minor pathway of cysteine catabolism: transamination and transsulfuration. Formation of thiosulfate.

## Sulfite Oxidase Deficiency

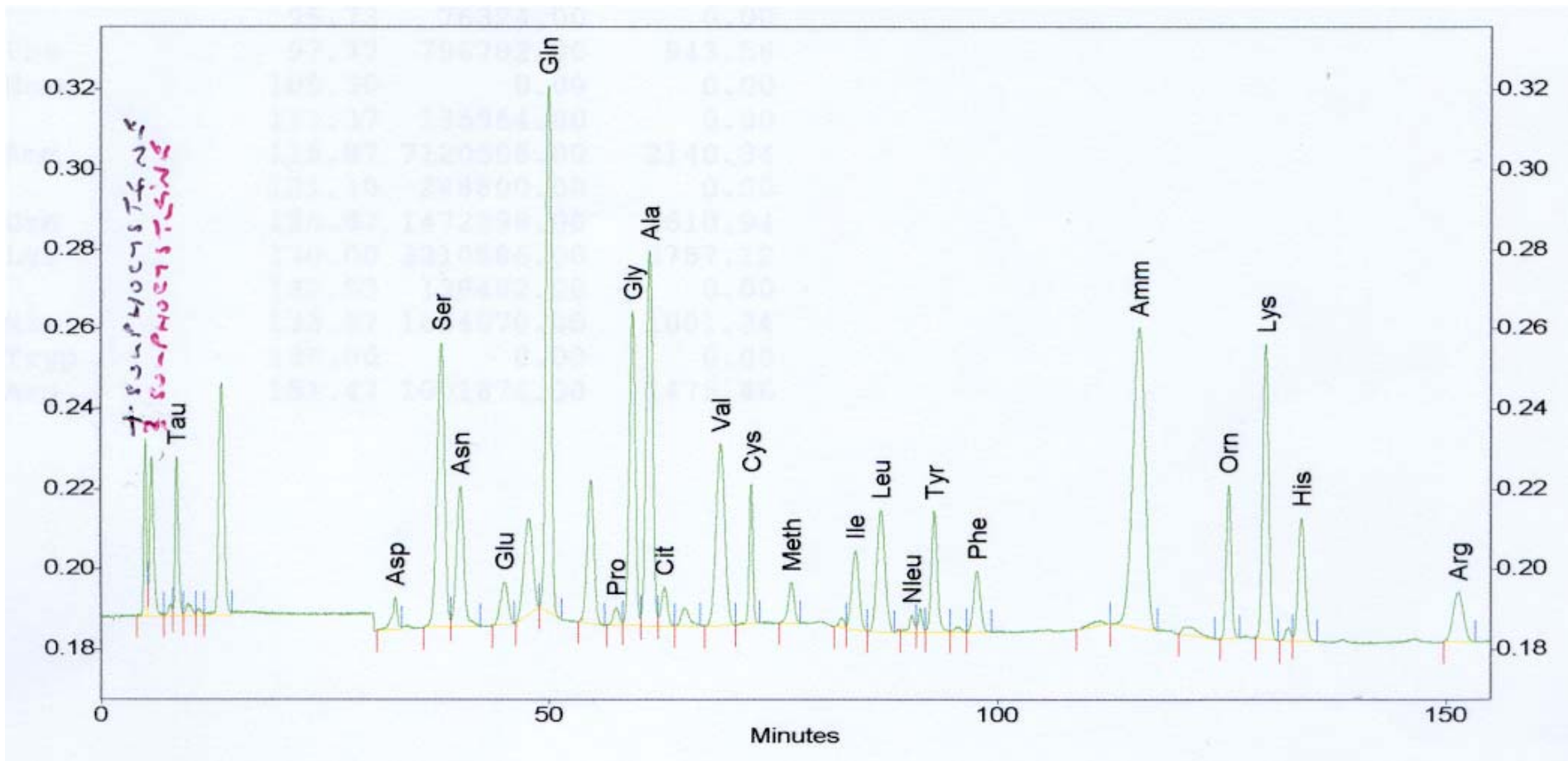
*Figs from Johnson and Wadman, 7th Edition, Scriver, MMBID*



# 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

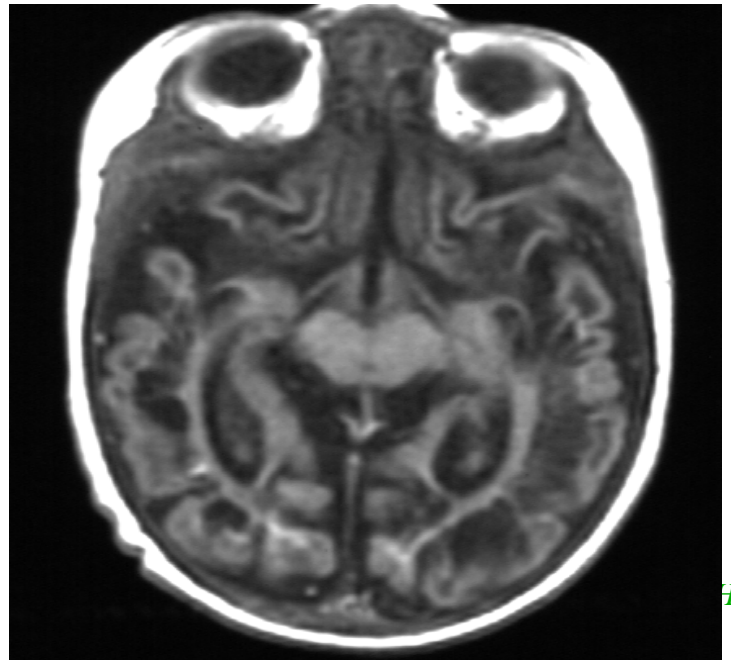
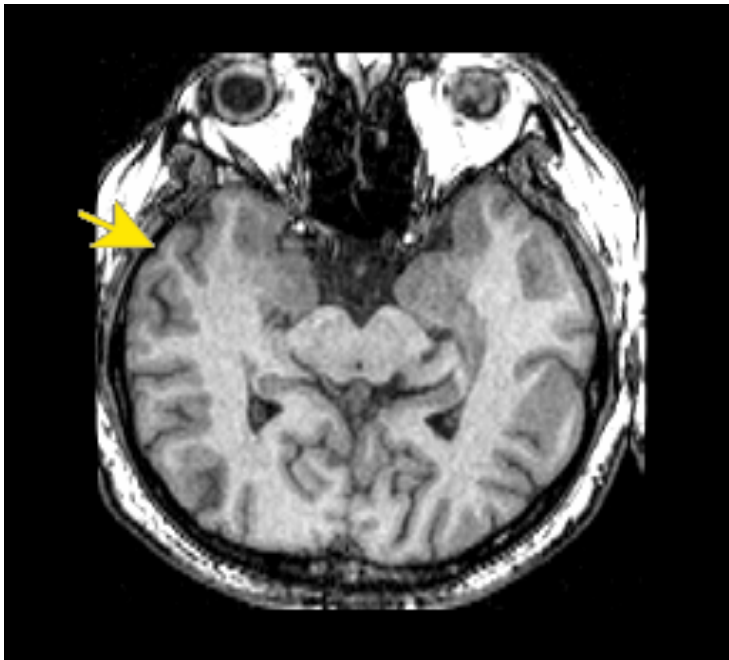
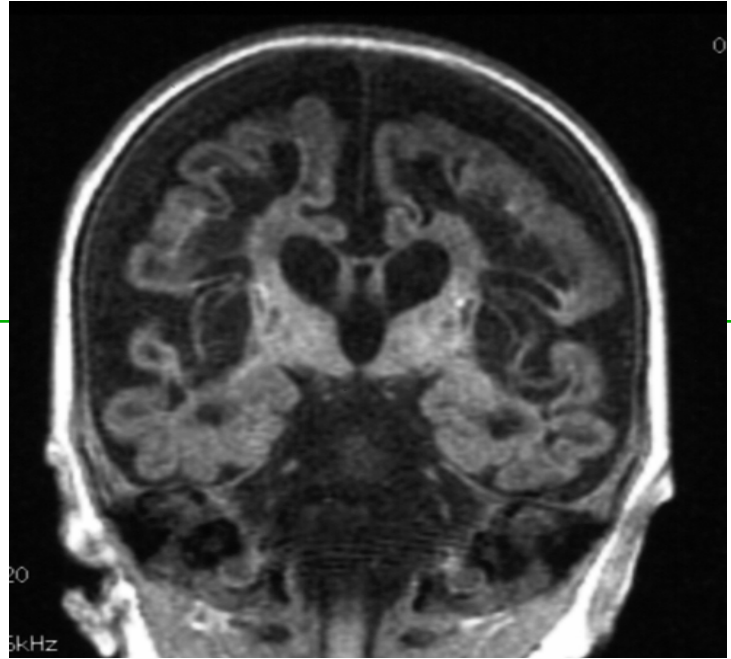
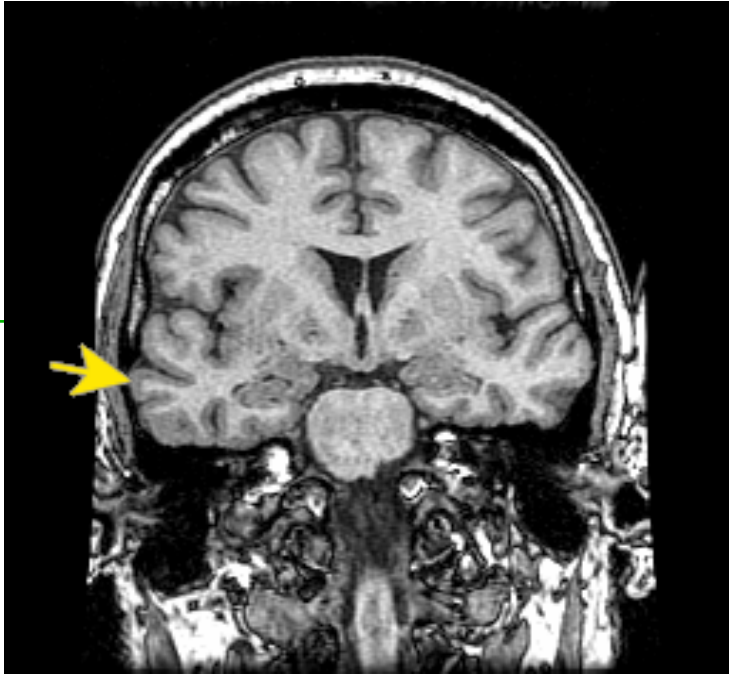
---

- 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

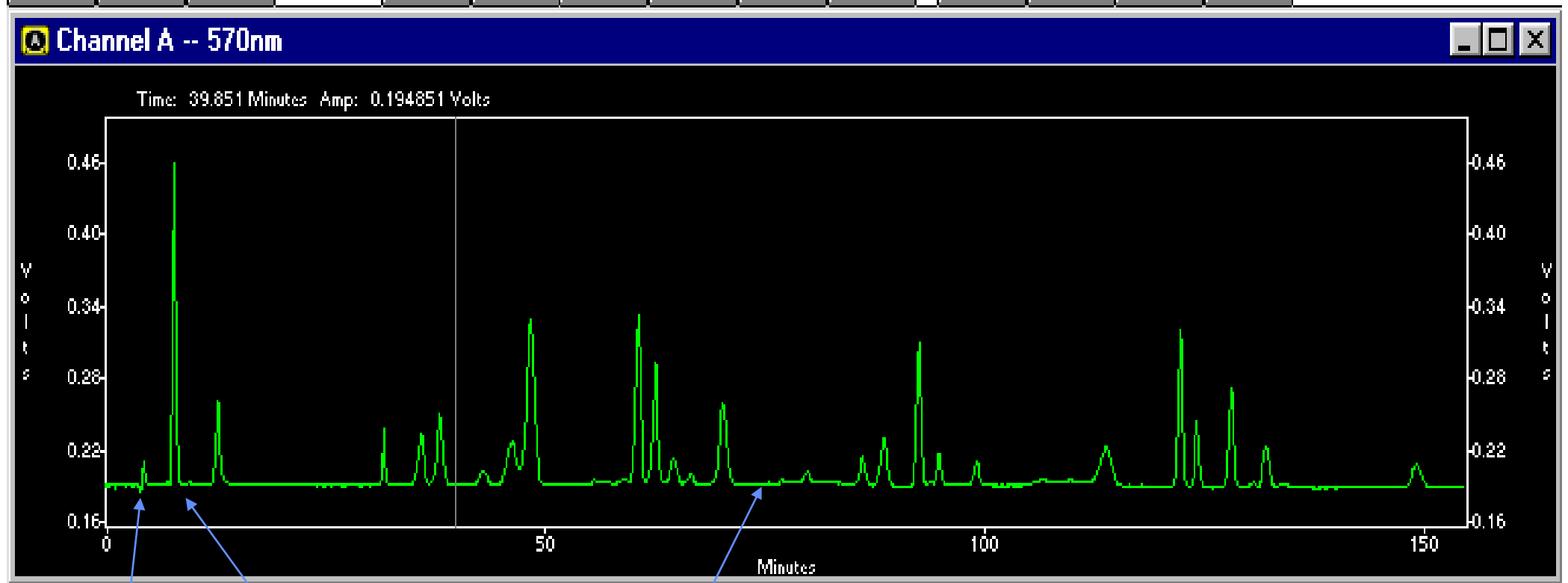
---

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



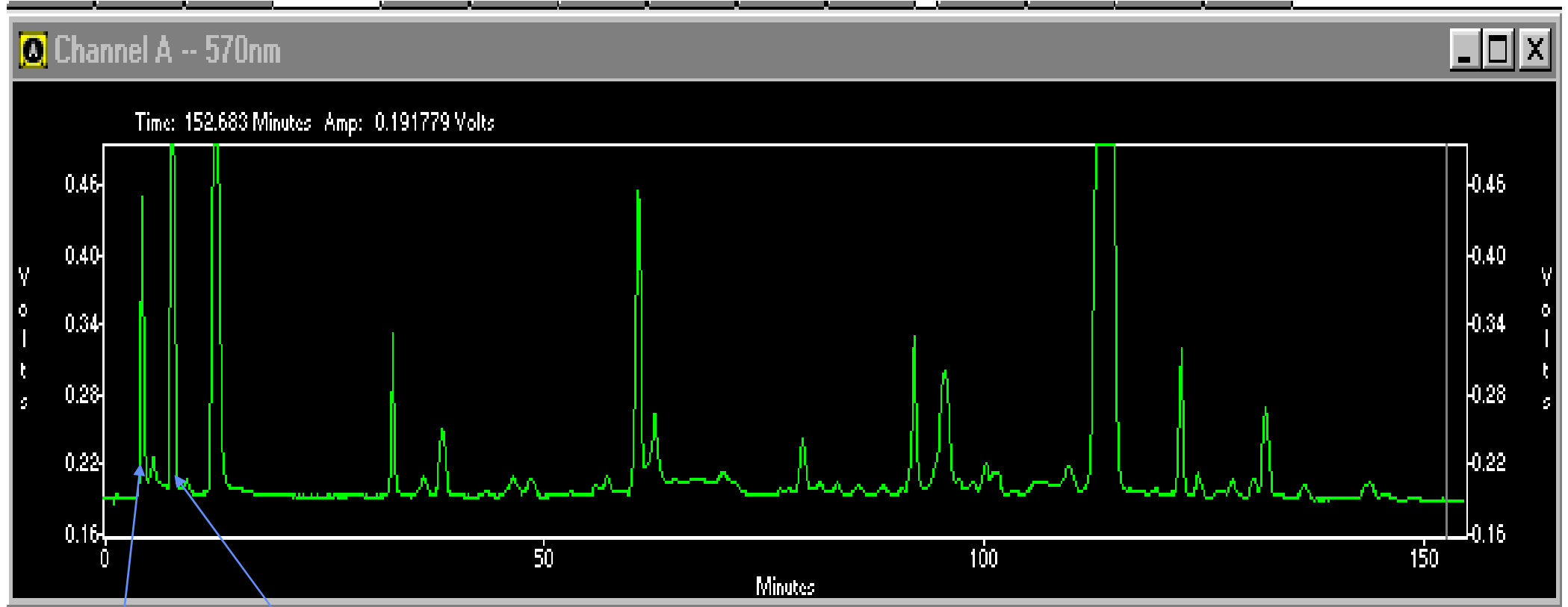
◆ sulphocysteine

◆ taurine

◆ no cystine

◆ Total plasma homocysteine undetectable.

# Urine



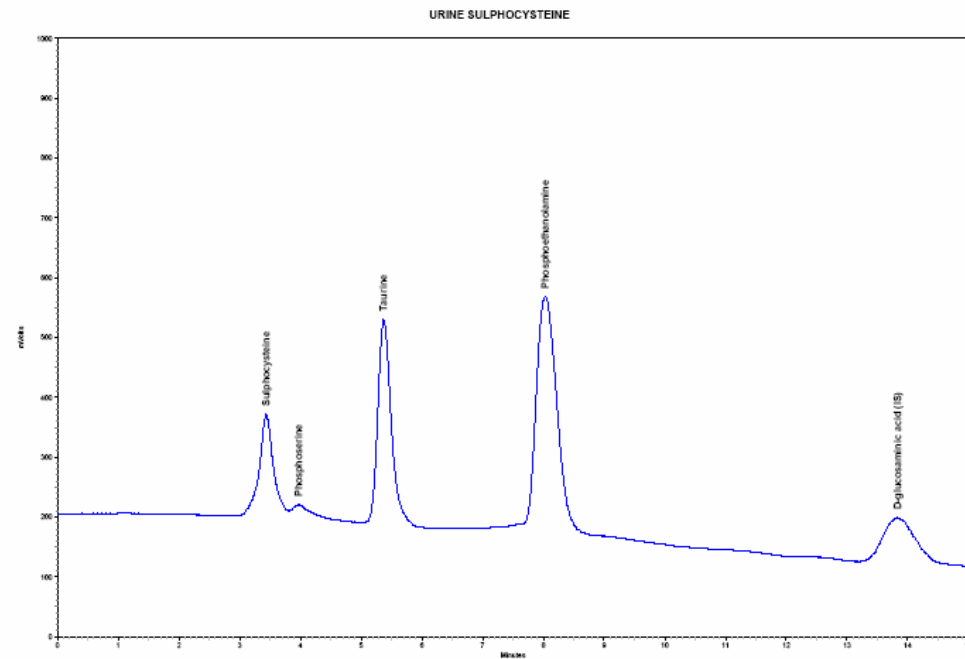
sulphocysteine

taurine

# Urine; Case 3, 2005

## New short programme for Biochrom

---



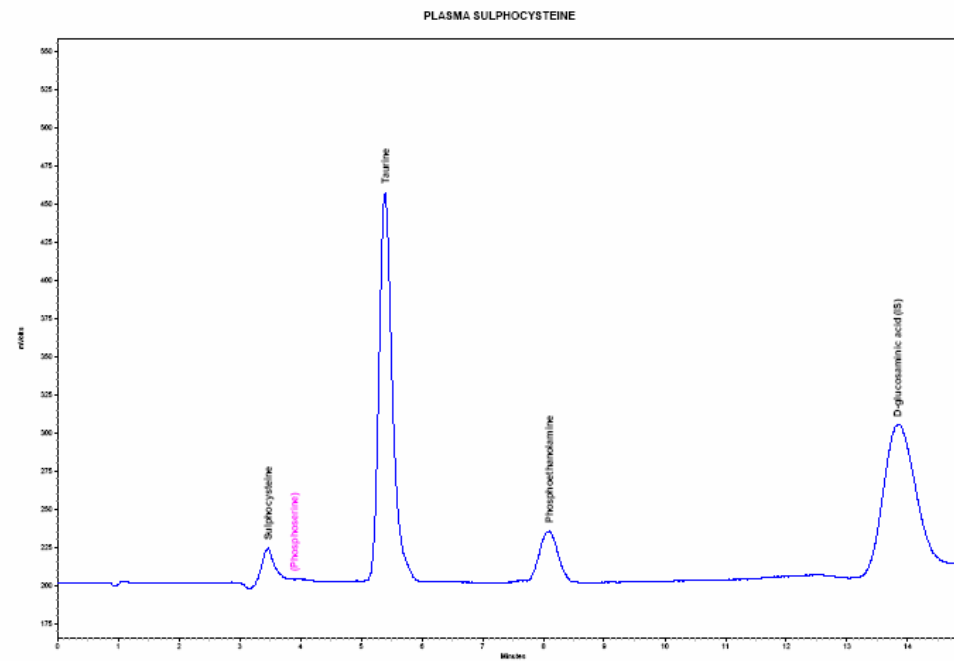
Ref: Applications note Bio30.4 - Rapid analysis of sulphocysteine

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

# Plasma; 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