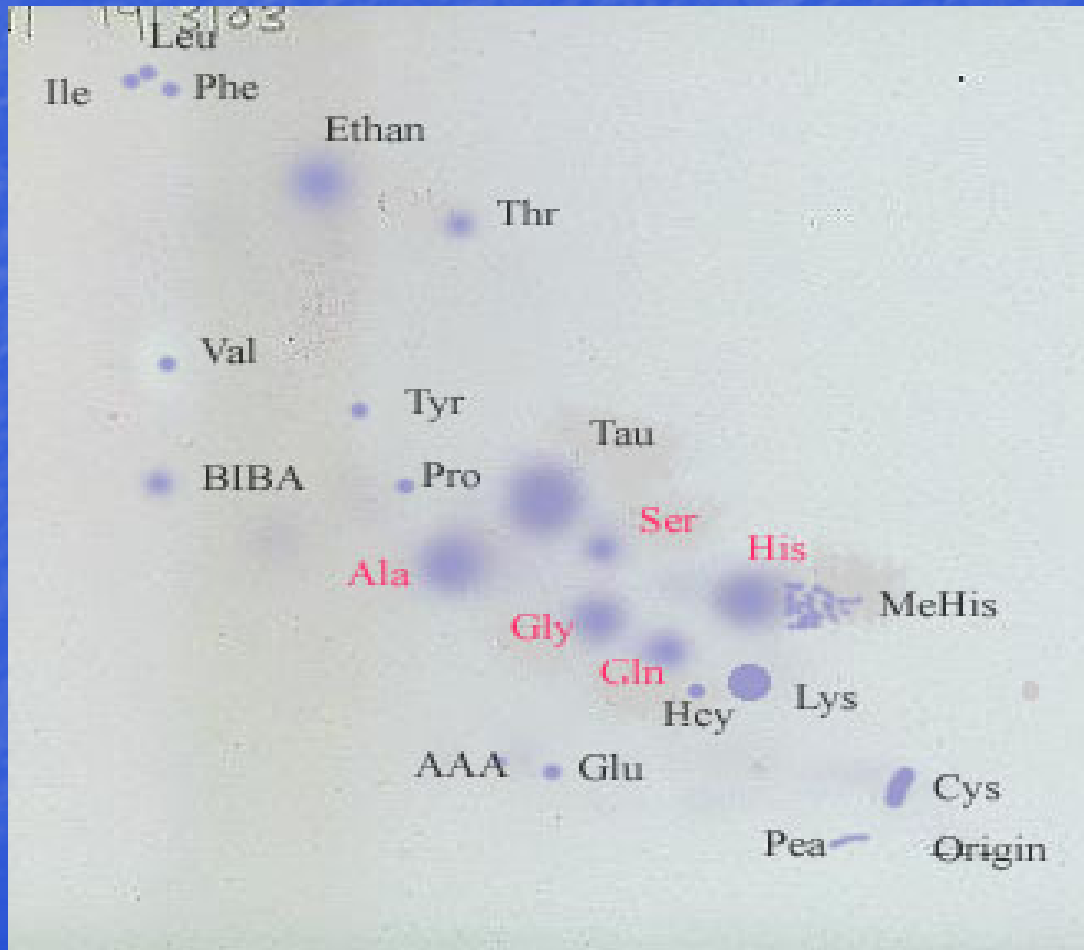




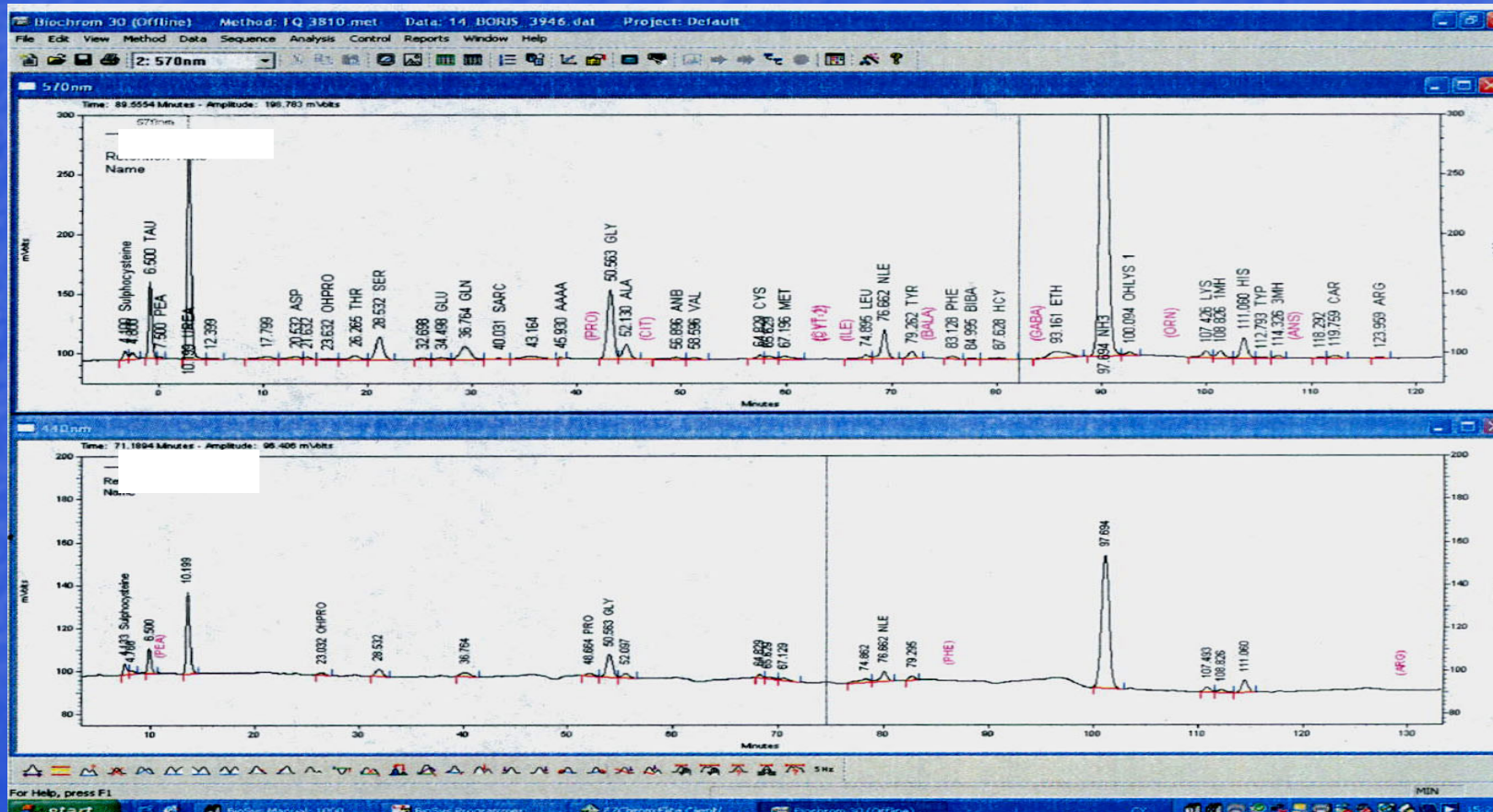
Problems with the Detection of Uncommon Amino Acids using the Biochrom 30

Jenny Watkinson
Sheffield Children's Hospital

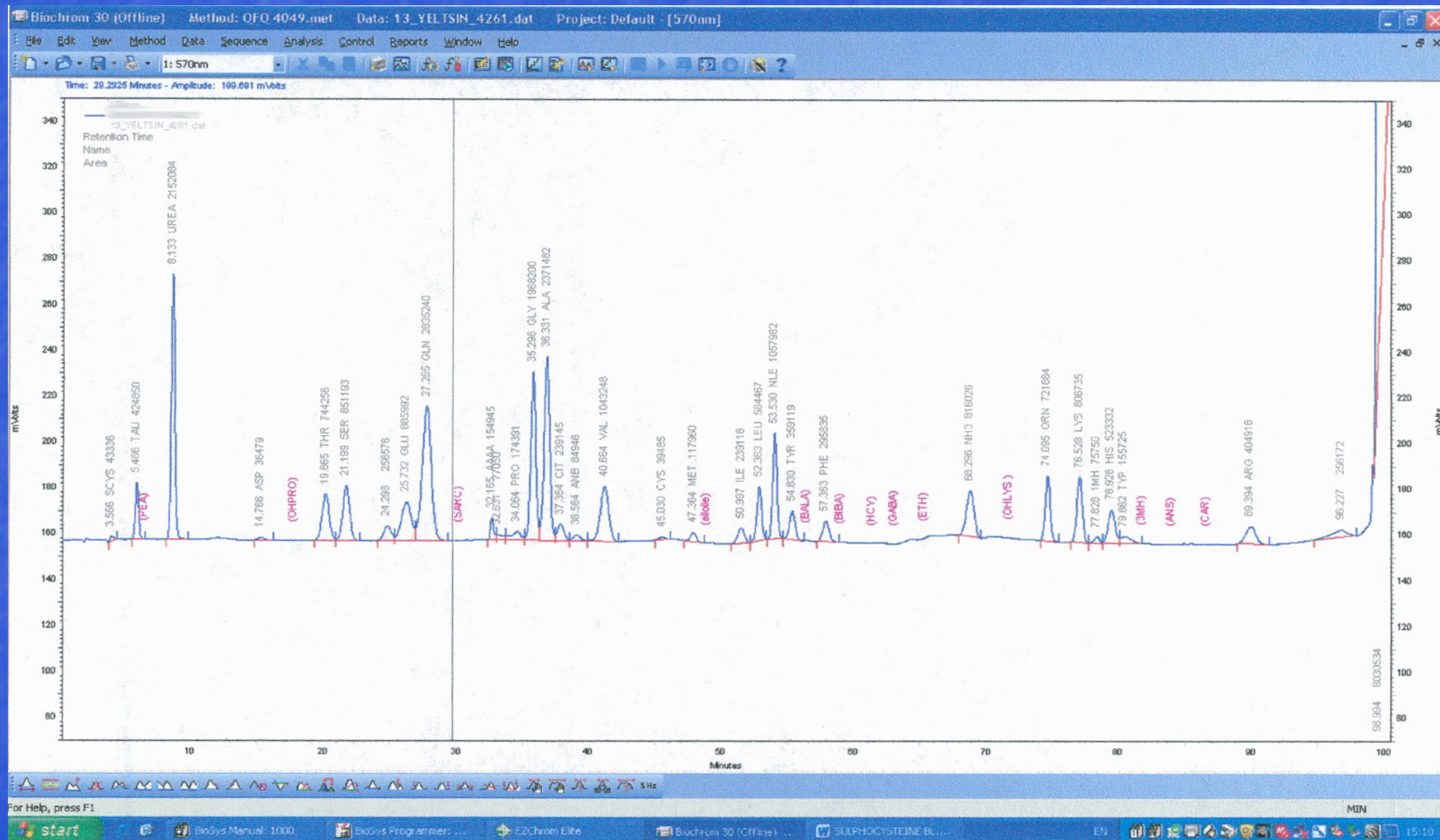
TLC Map



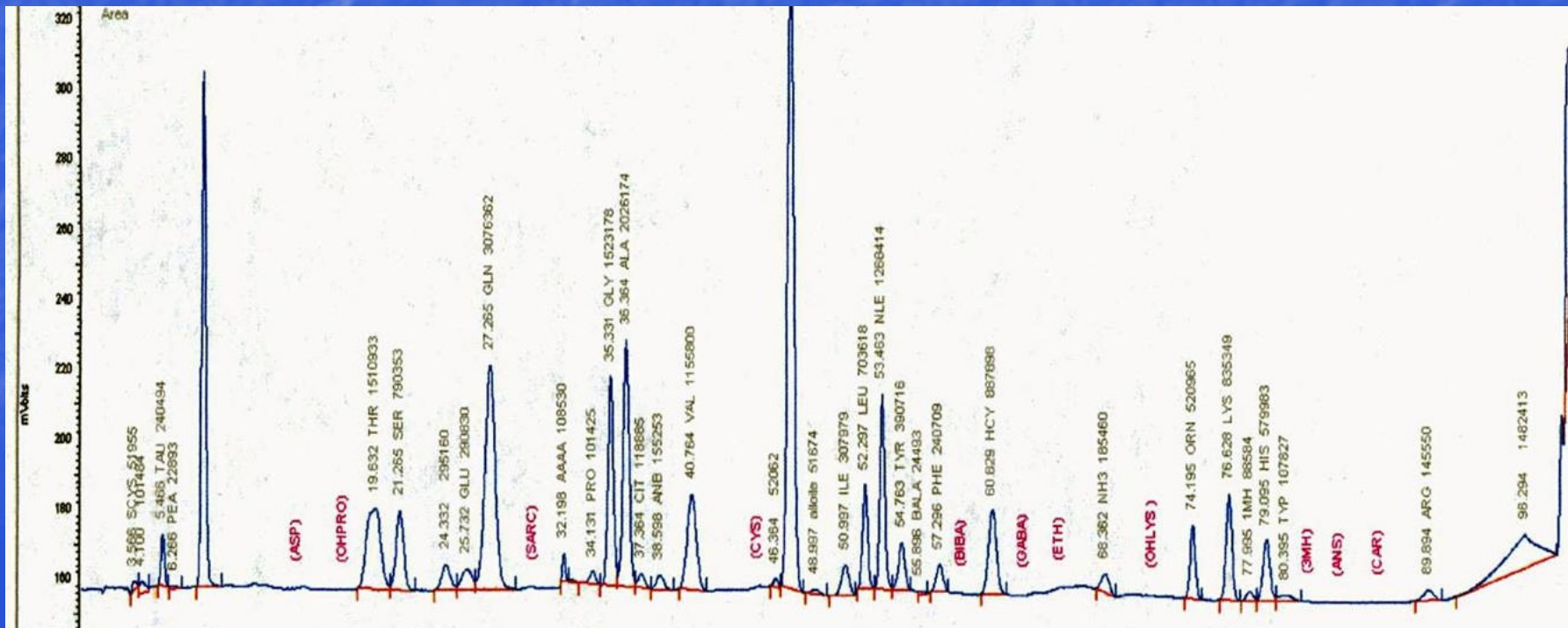
Normal Urine



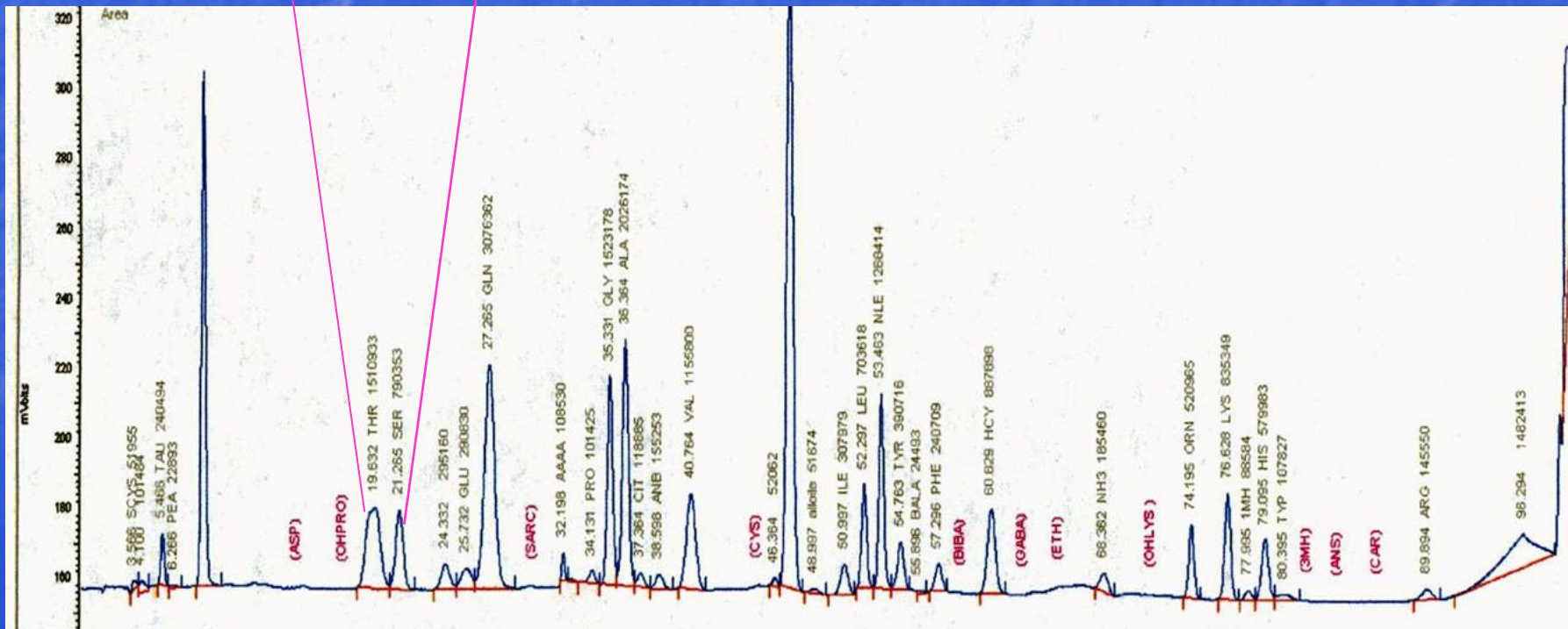
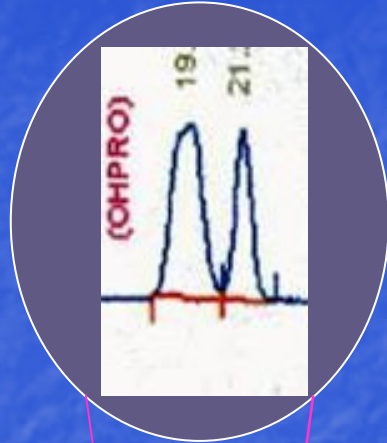
Normal Plasma



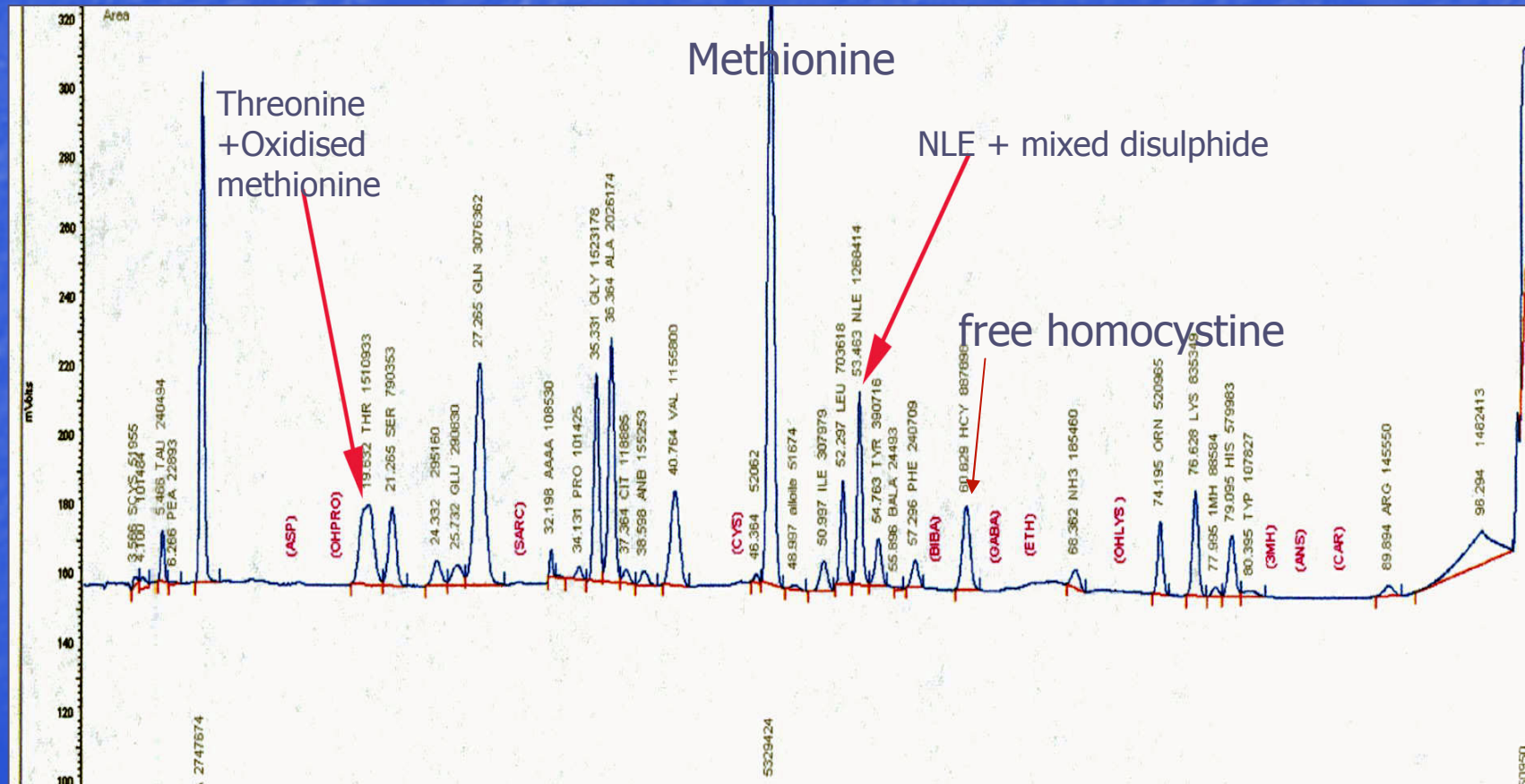
Plasma with abnormalities



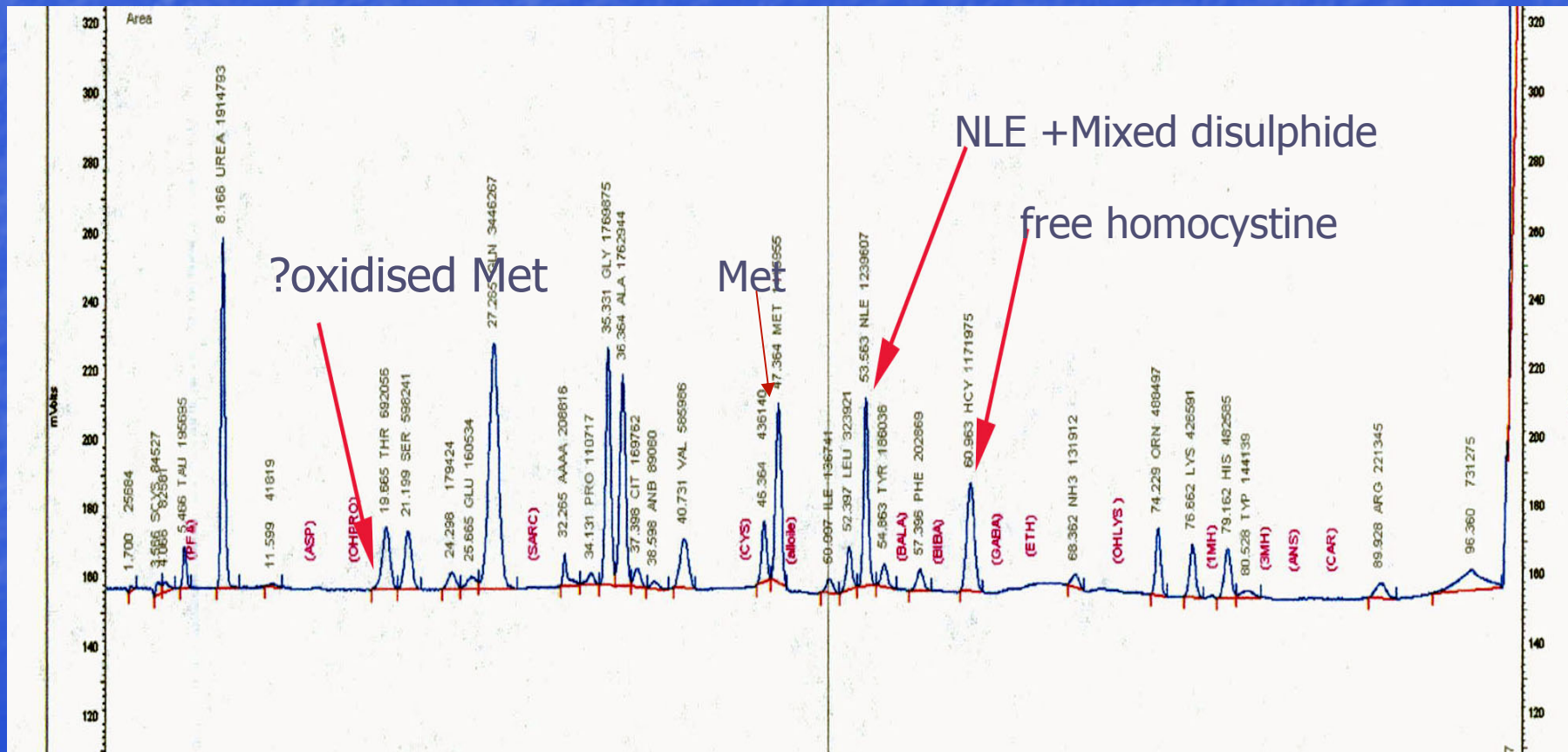
Plasma with abnormalities



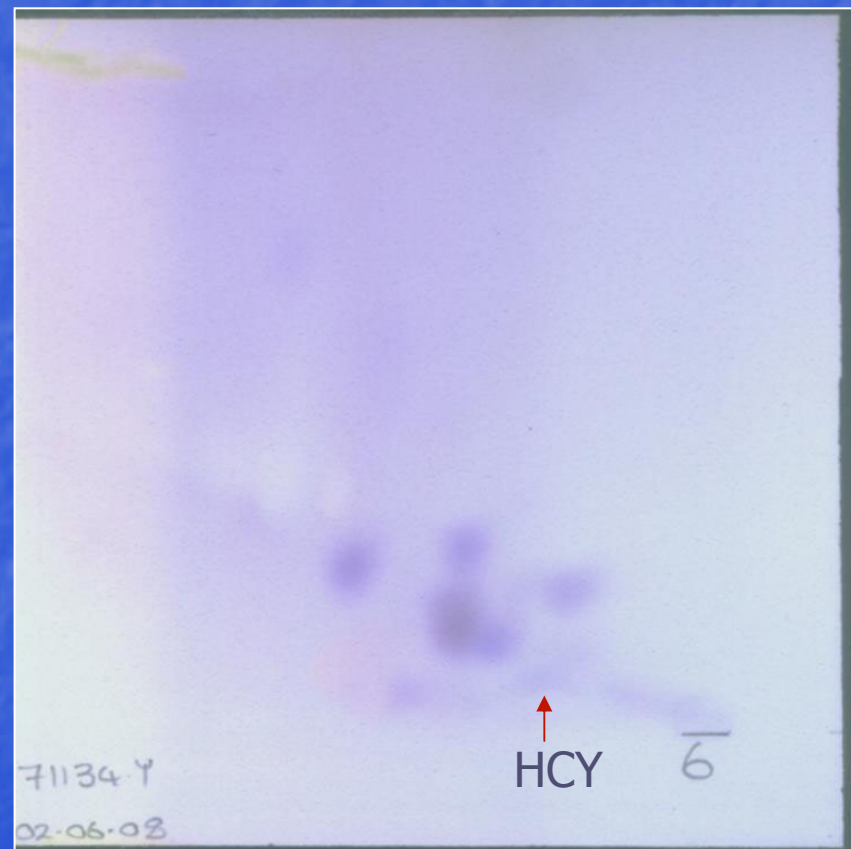
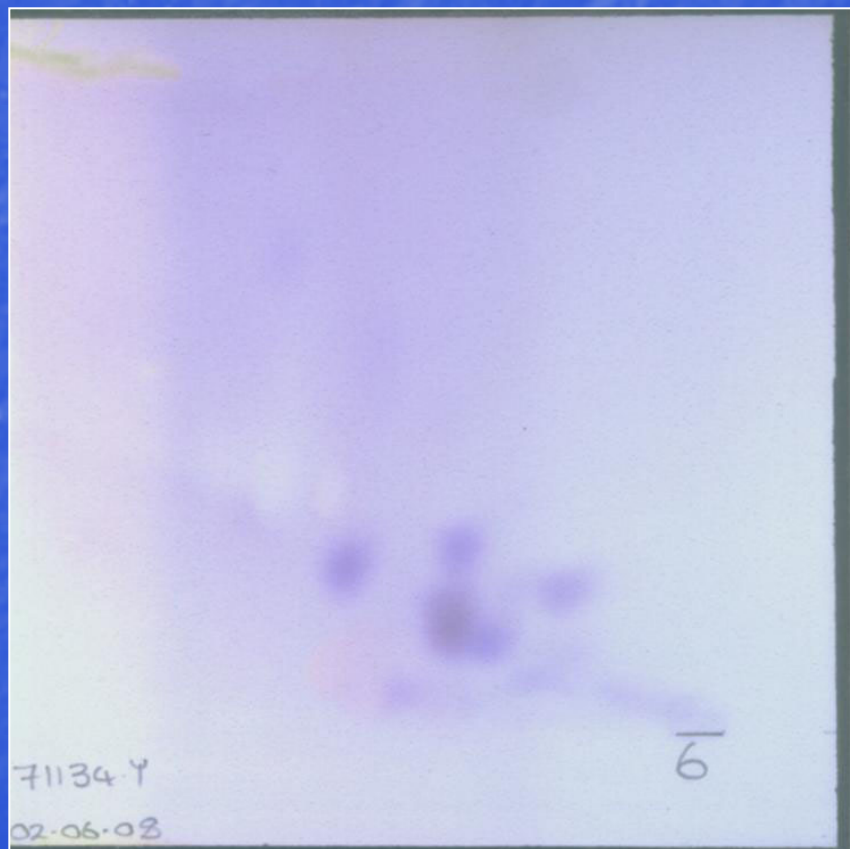
Patient with Homocystinuria



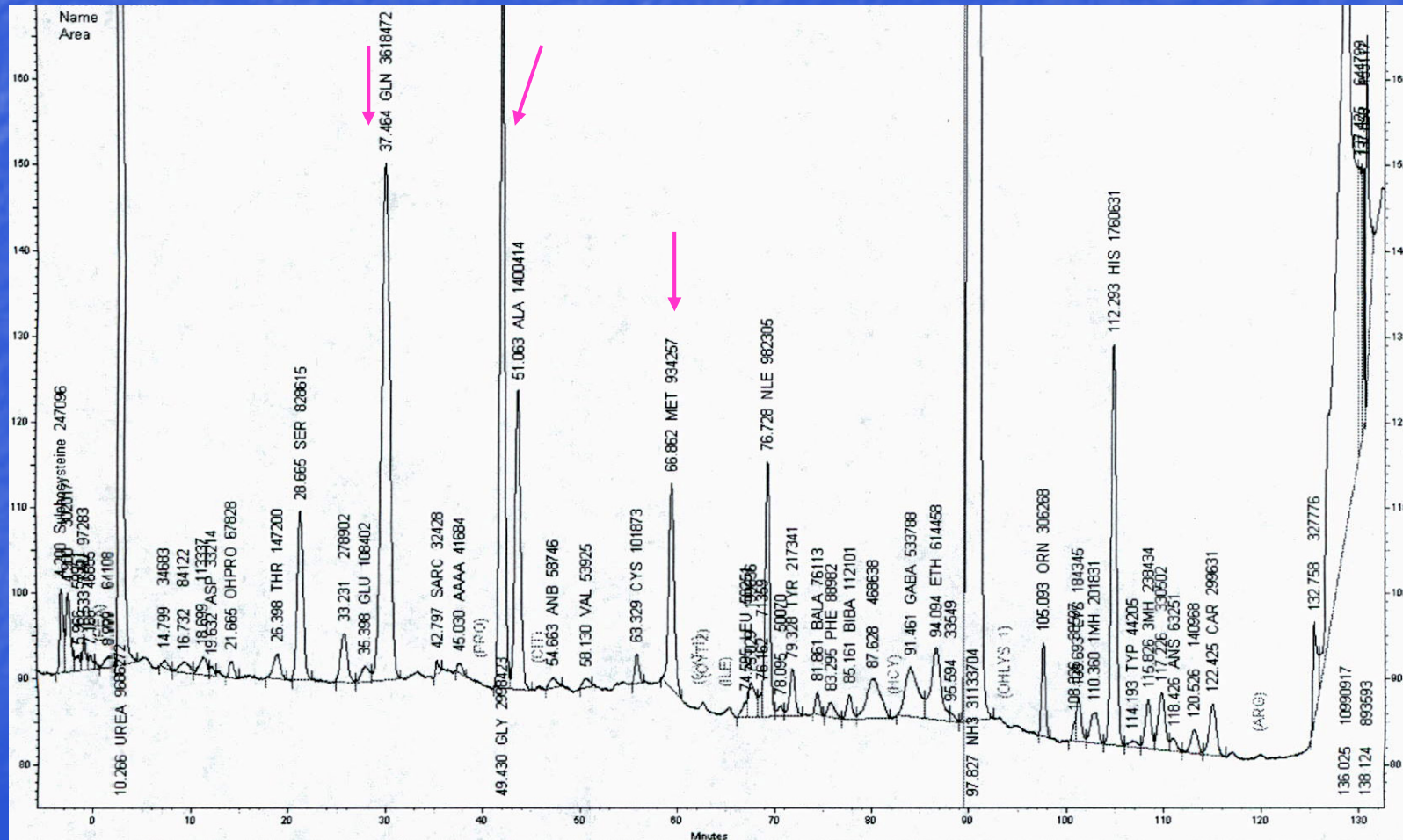
Sibling of previous patient



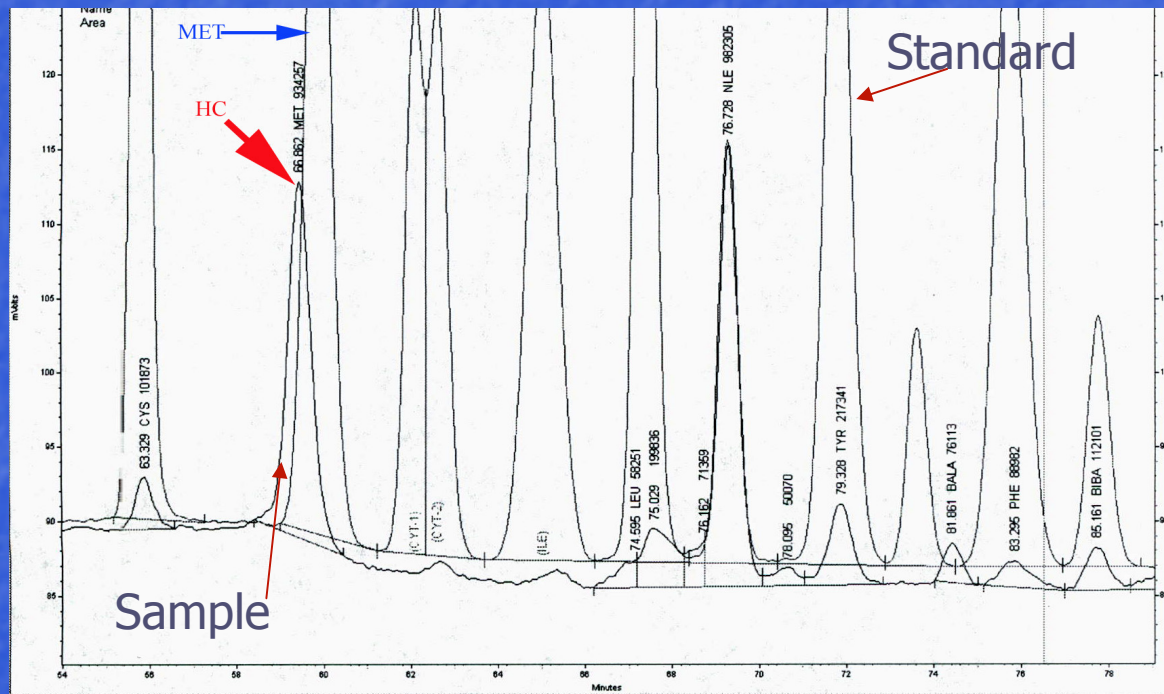
Is TLC useful ?



Which are the abnormal peaks?

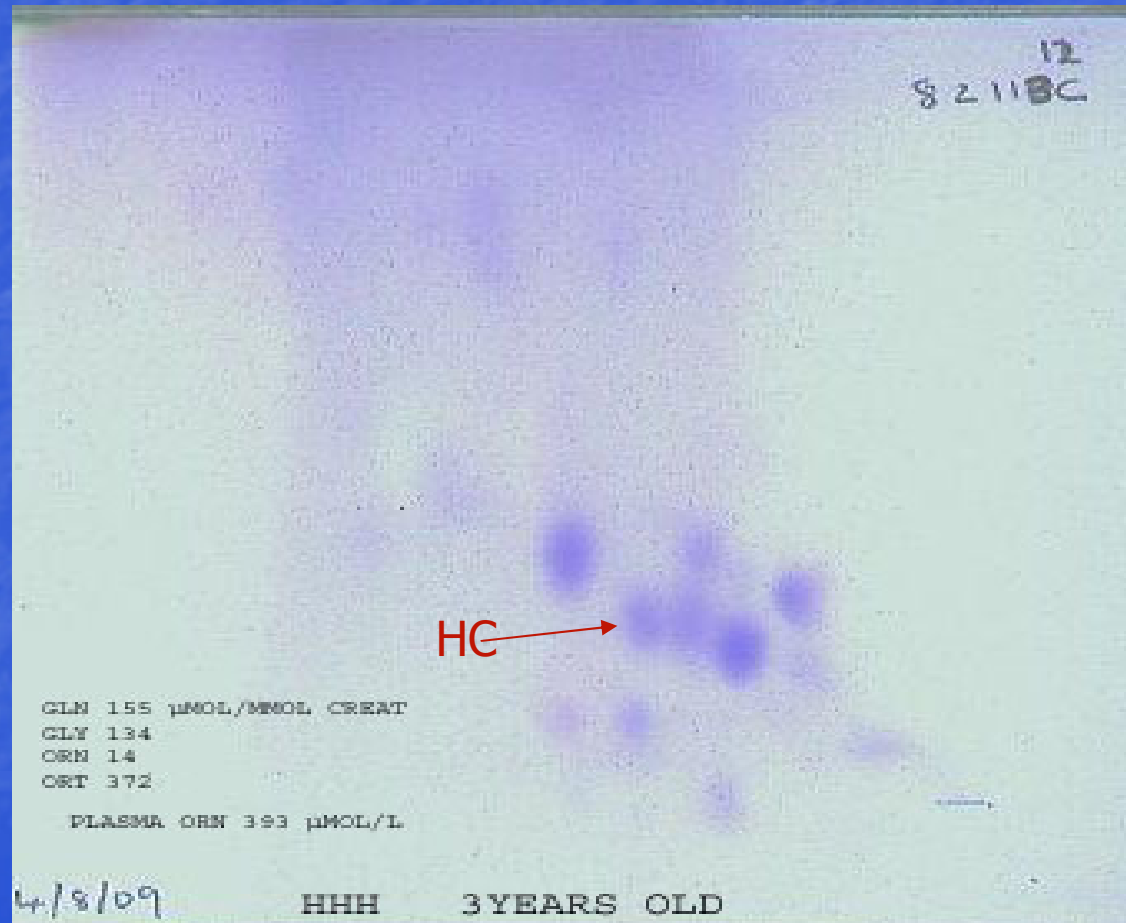


Short Run on Biochrom 30

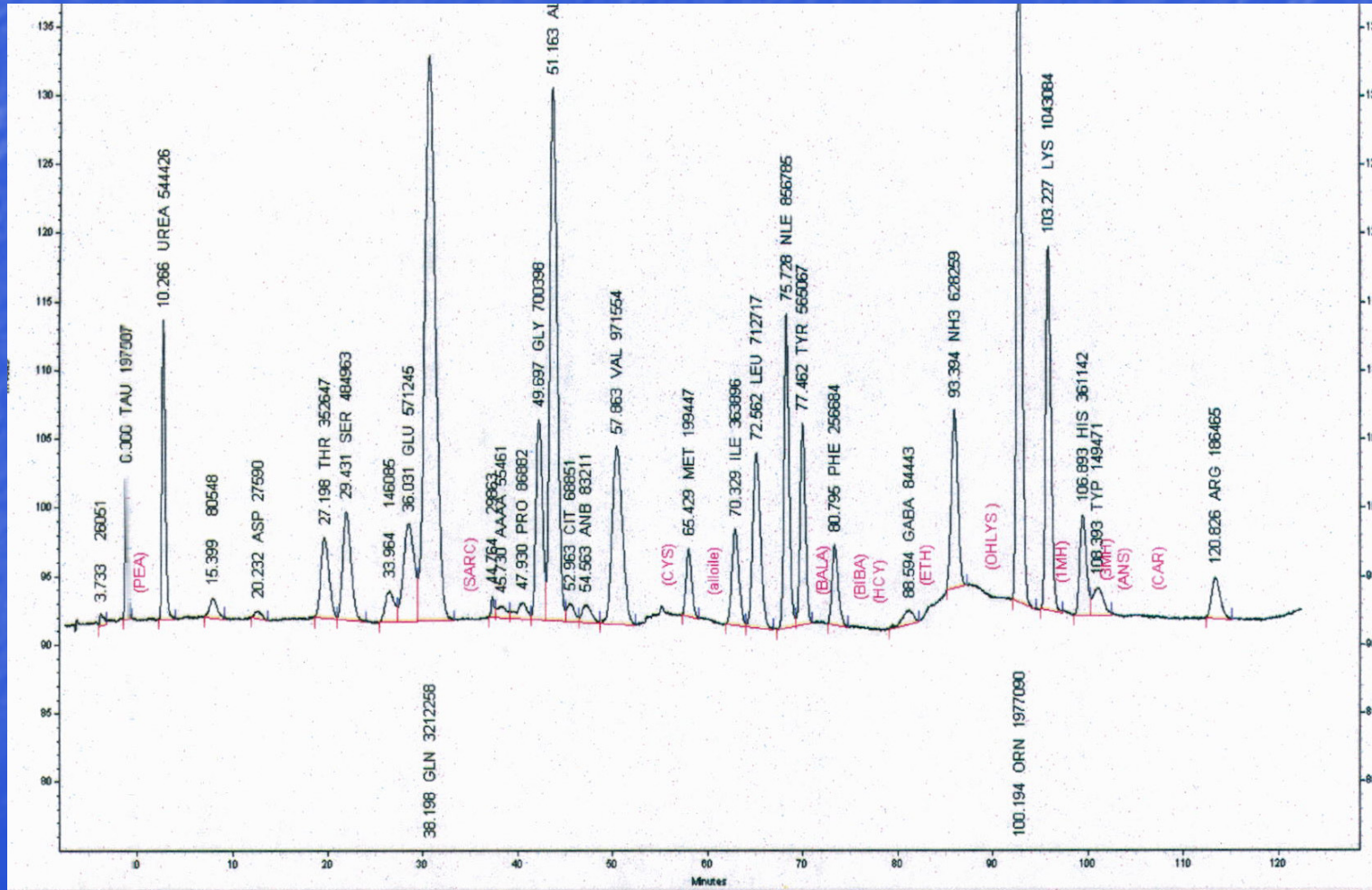


Overlay with Sigma standard

Is TLC USEFUL ?



Plasma on same patient



EZCHROM Urine trace

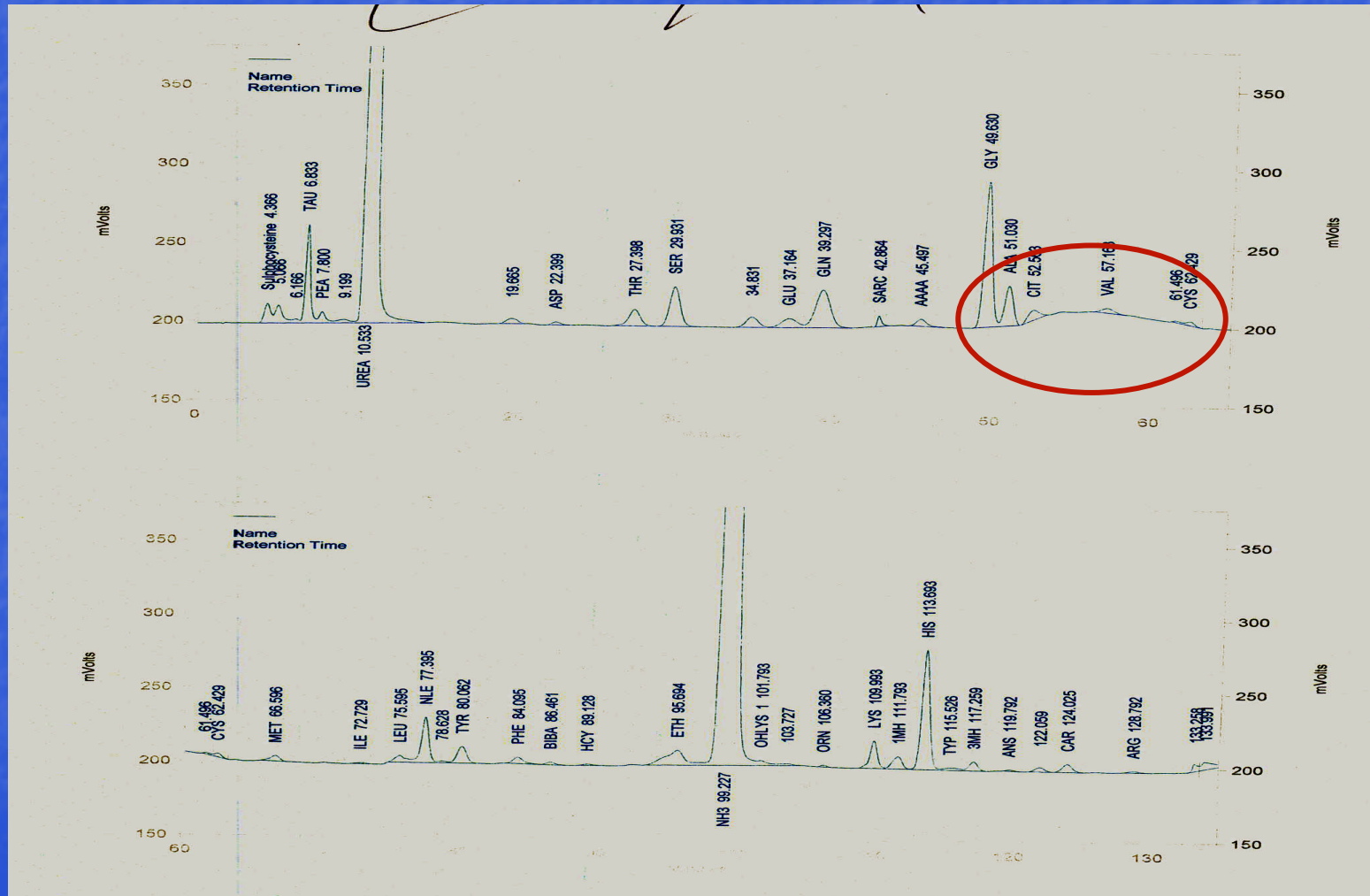
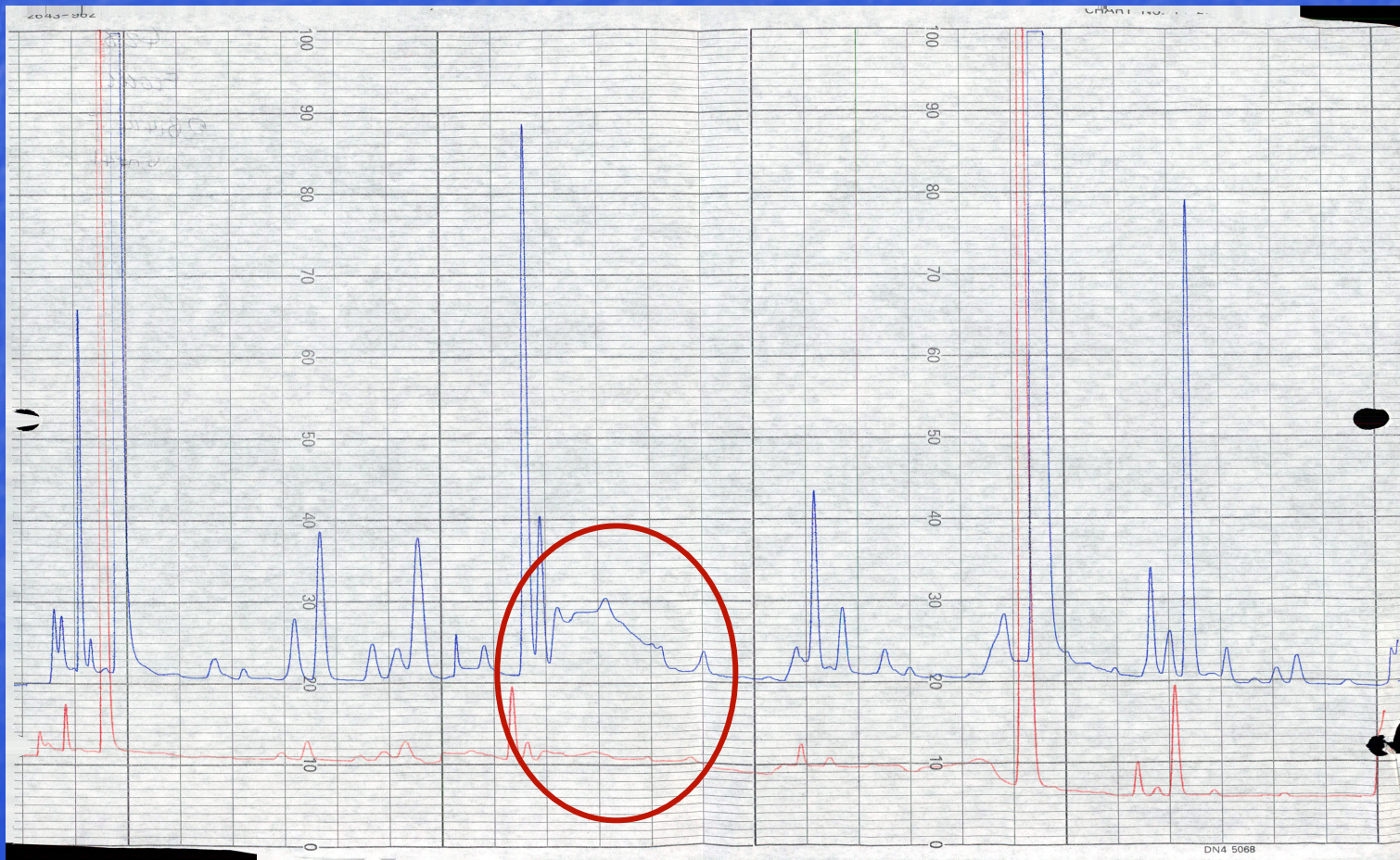
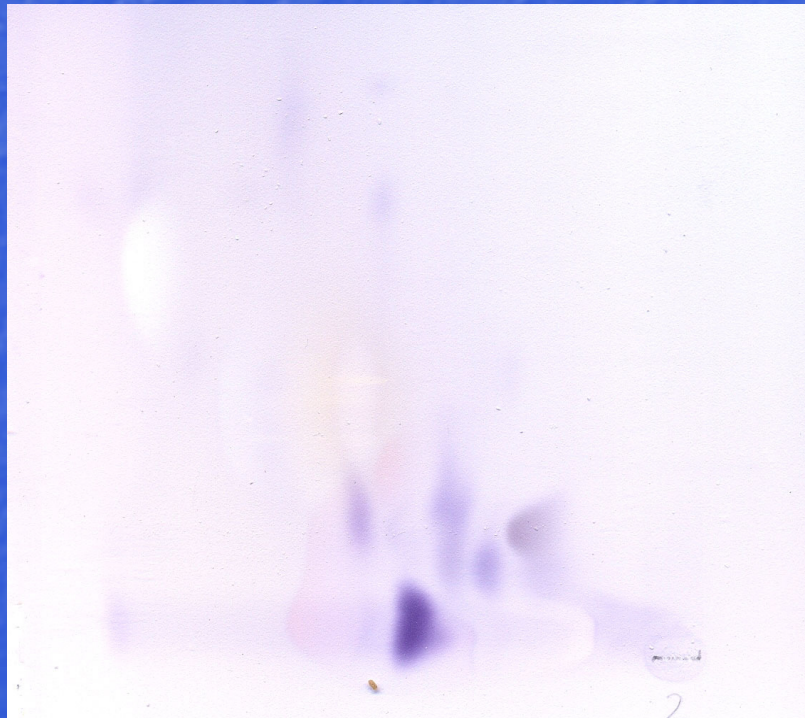


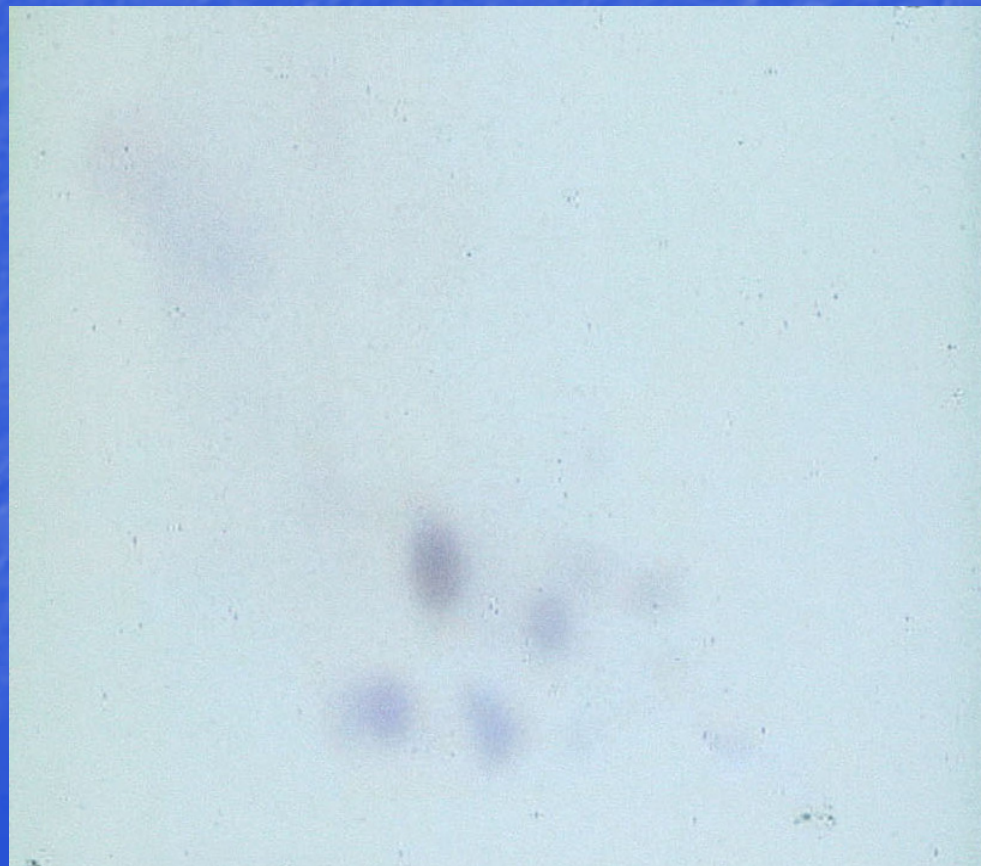
Chart Recorder Trace



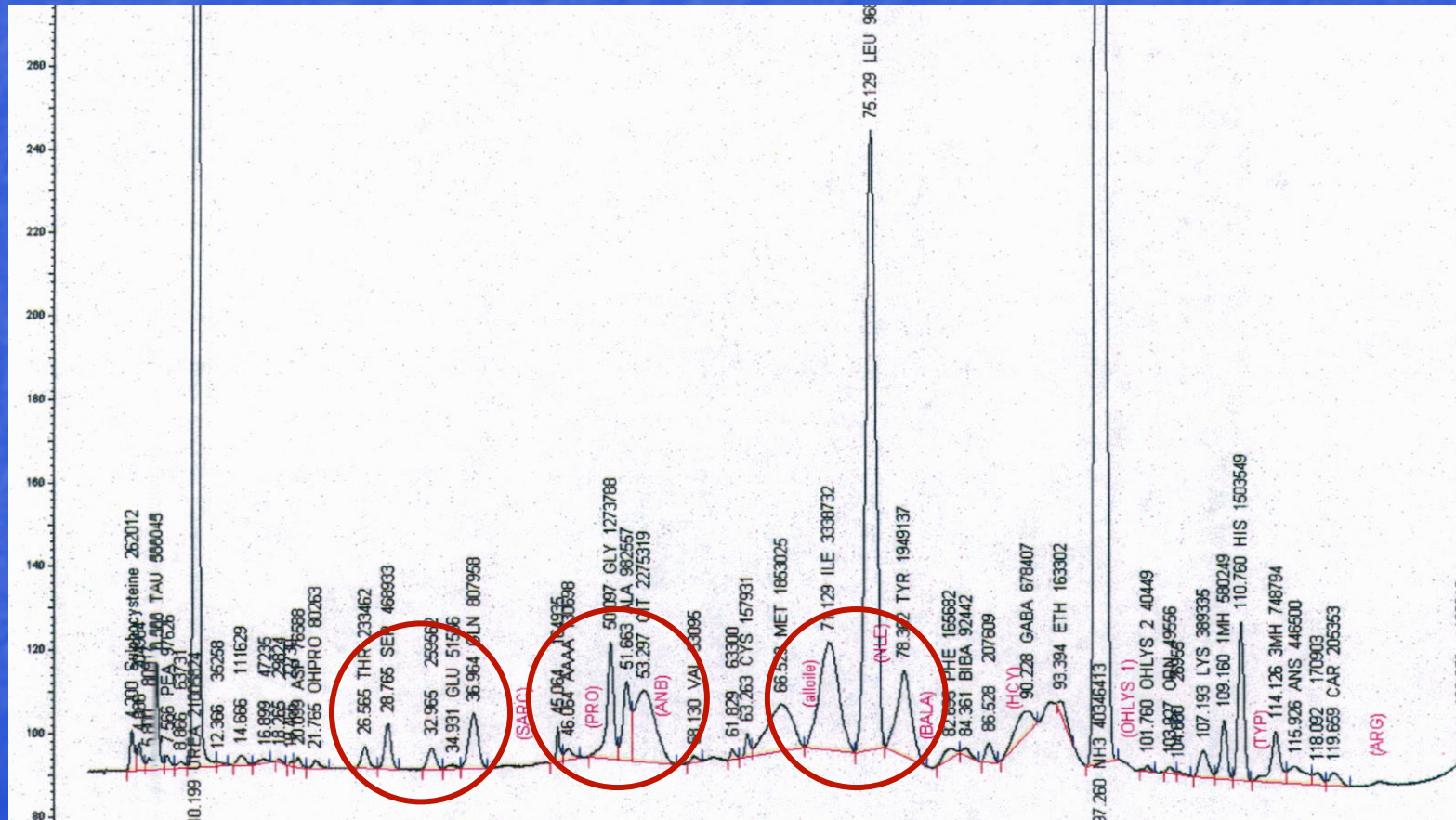
TLC



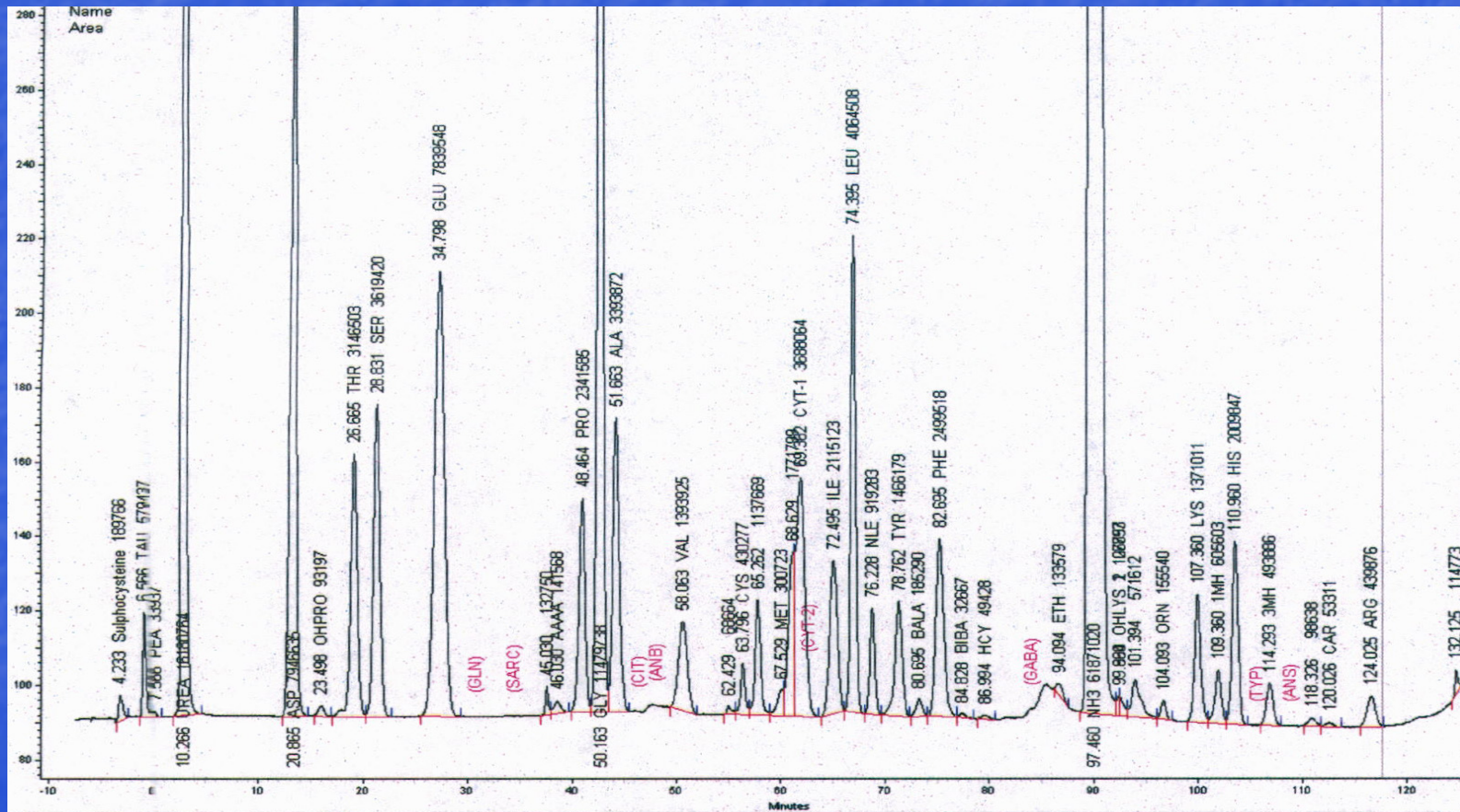
TLC - Deterioration??



?? Drugs or Deterioration?

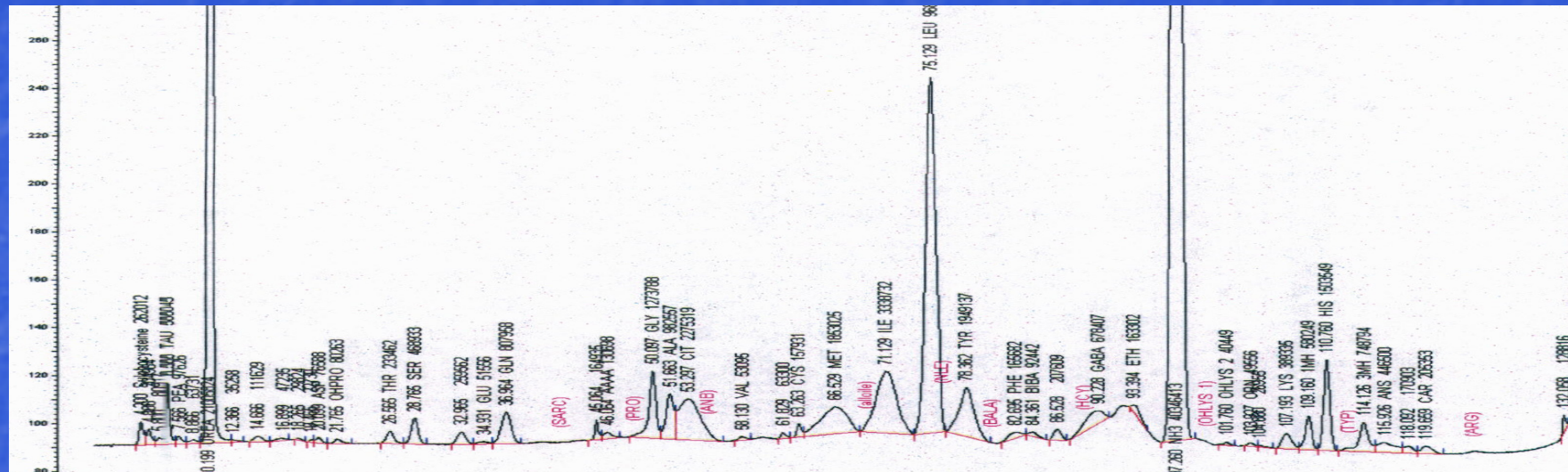


Same sample post hydrolysis

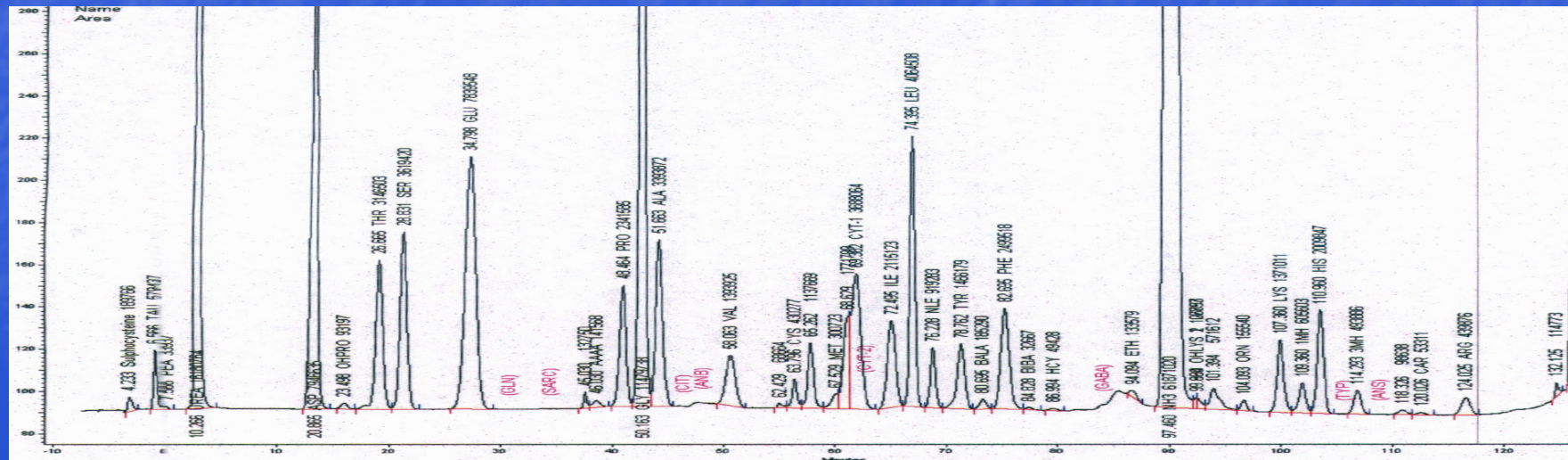


Prolidase Deficiency

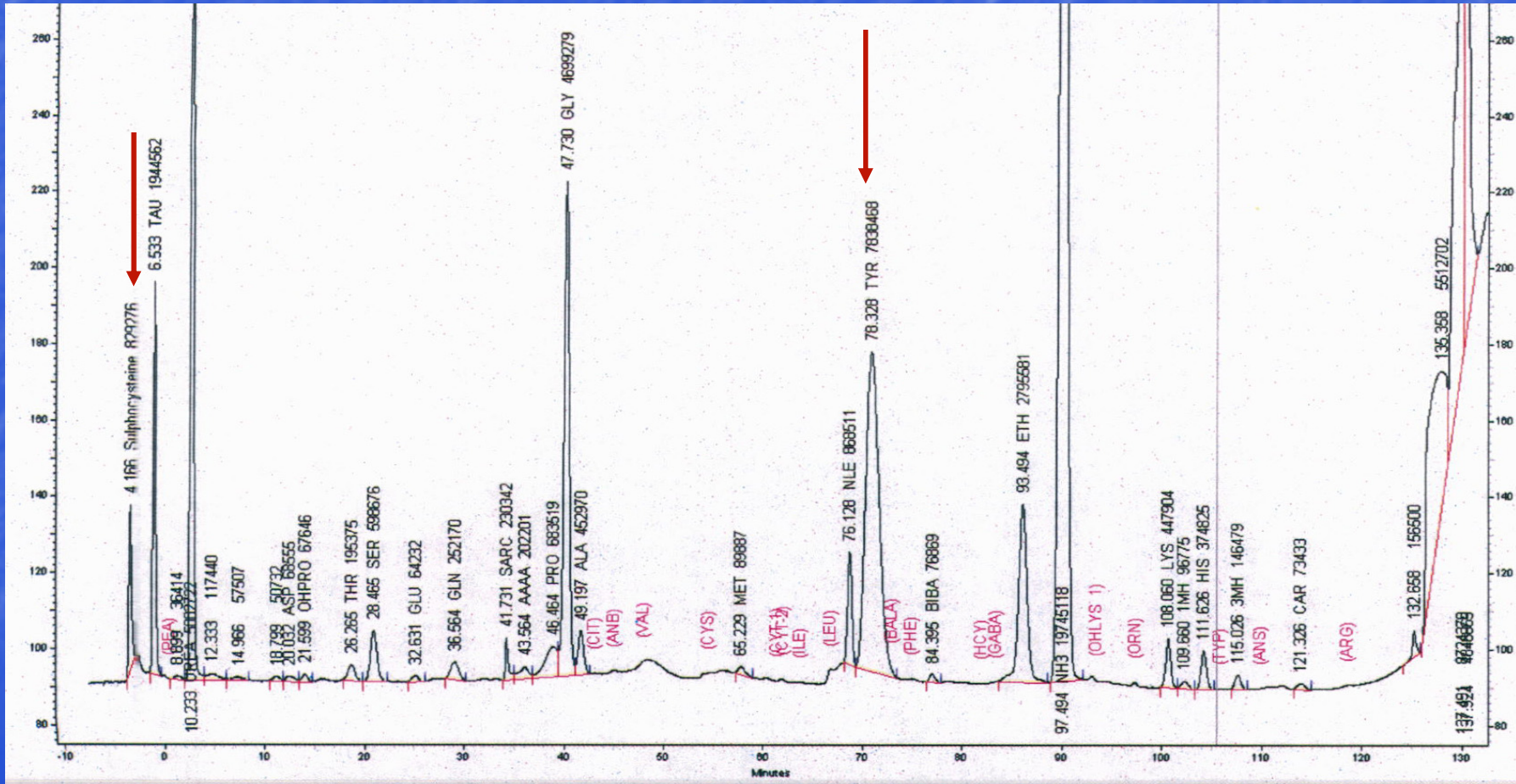
Original Sample:



Sample post hydrolysis:



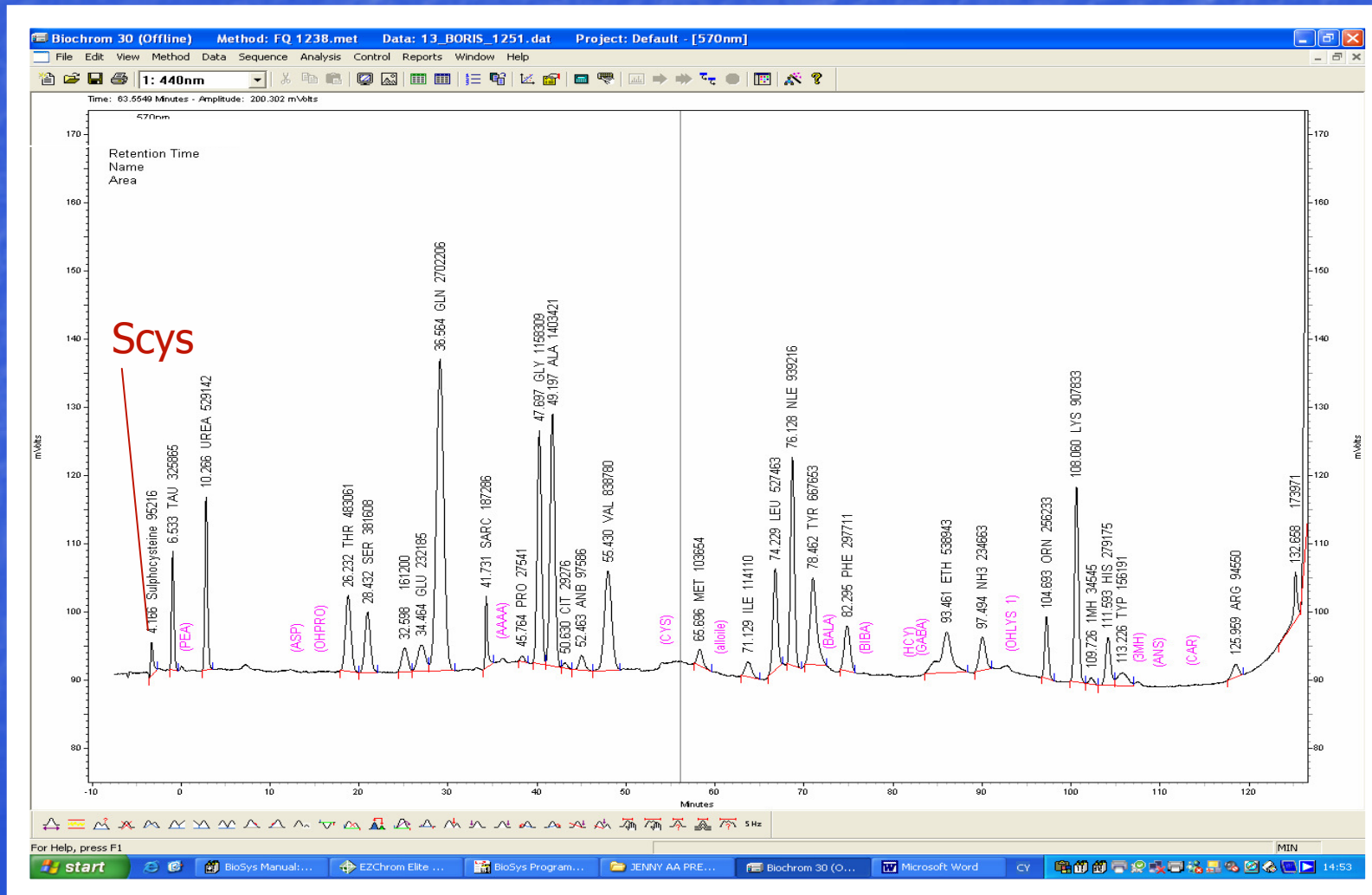
Urine: ? Drugs



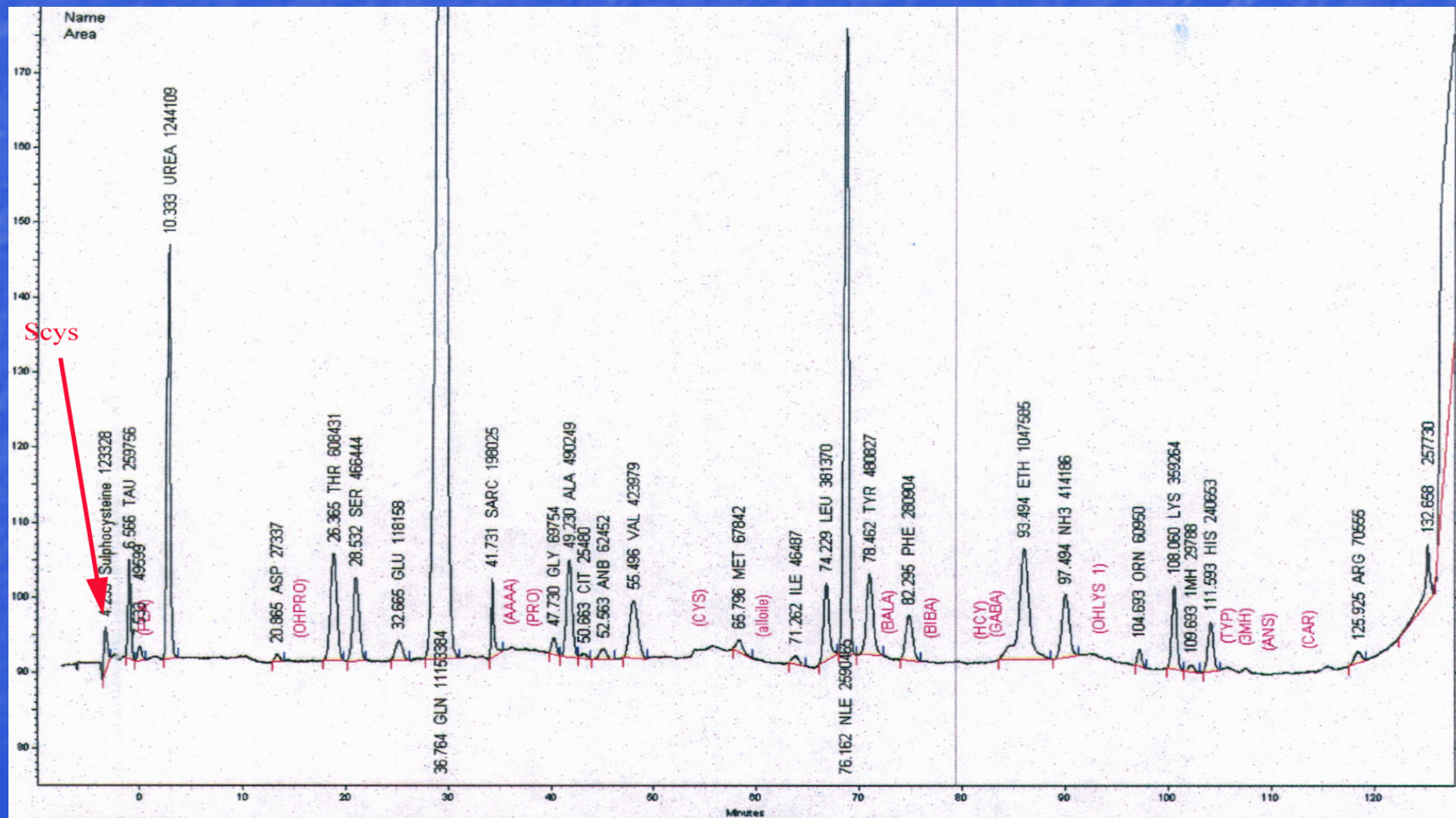
Urine Scys = 251 $\mu\text{mol}/\text{mmol}$ creatinine



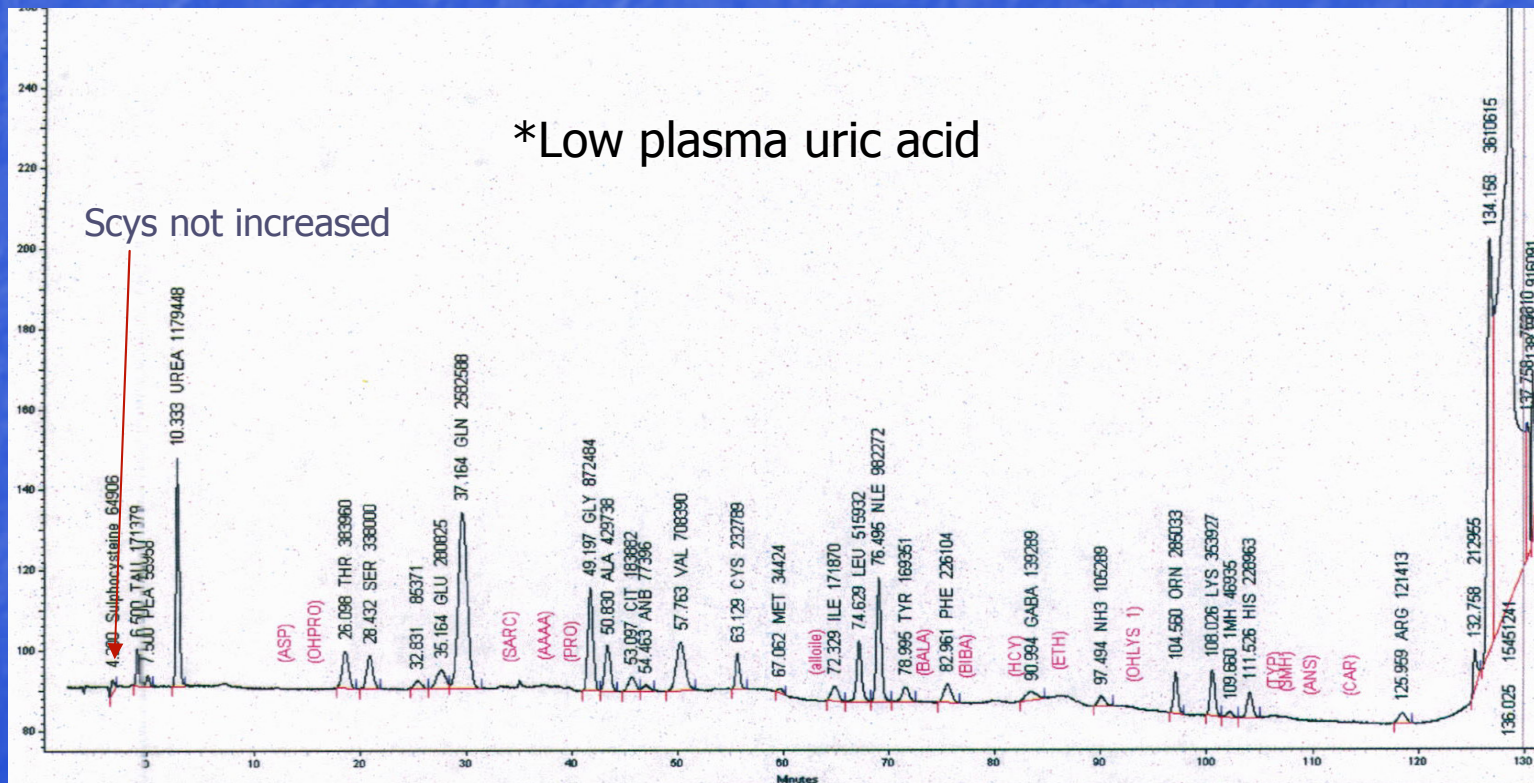
Plasma on same patient



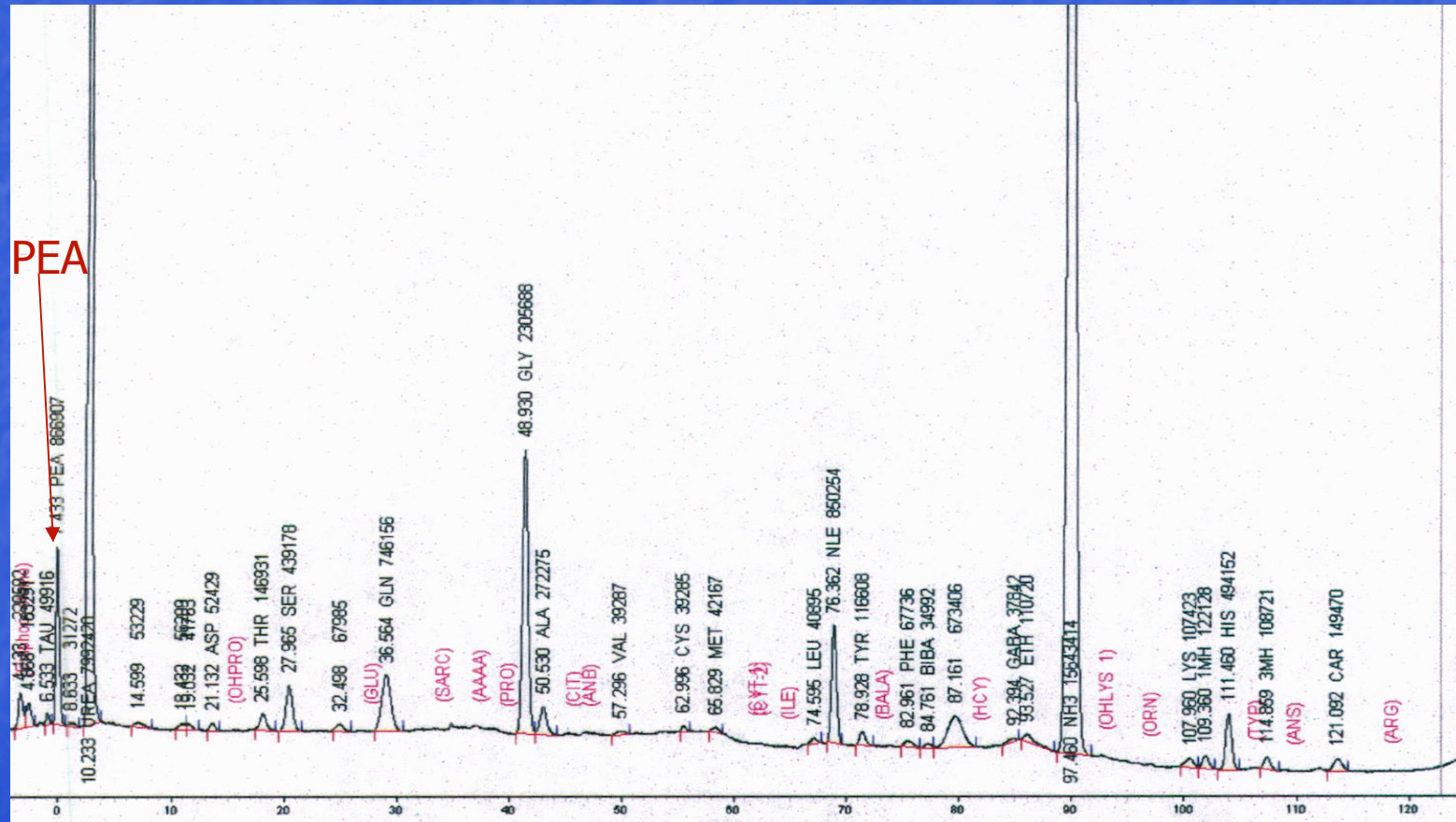
CSF sample same patient



Plasma on same patient, diagnosed with Molybdenum Cofactor Deficiency



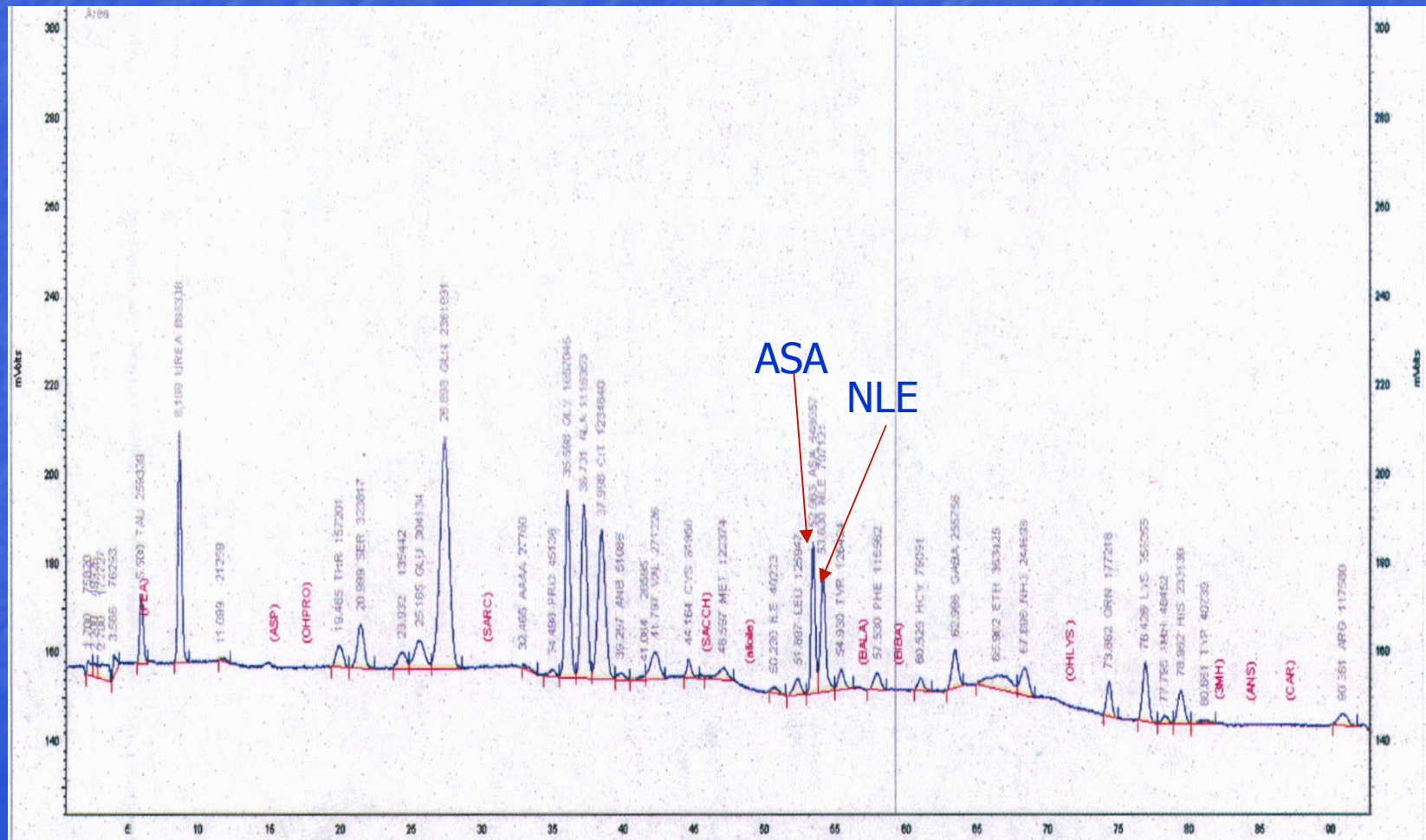
Urine from patient with hypophosphatasia



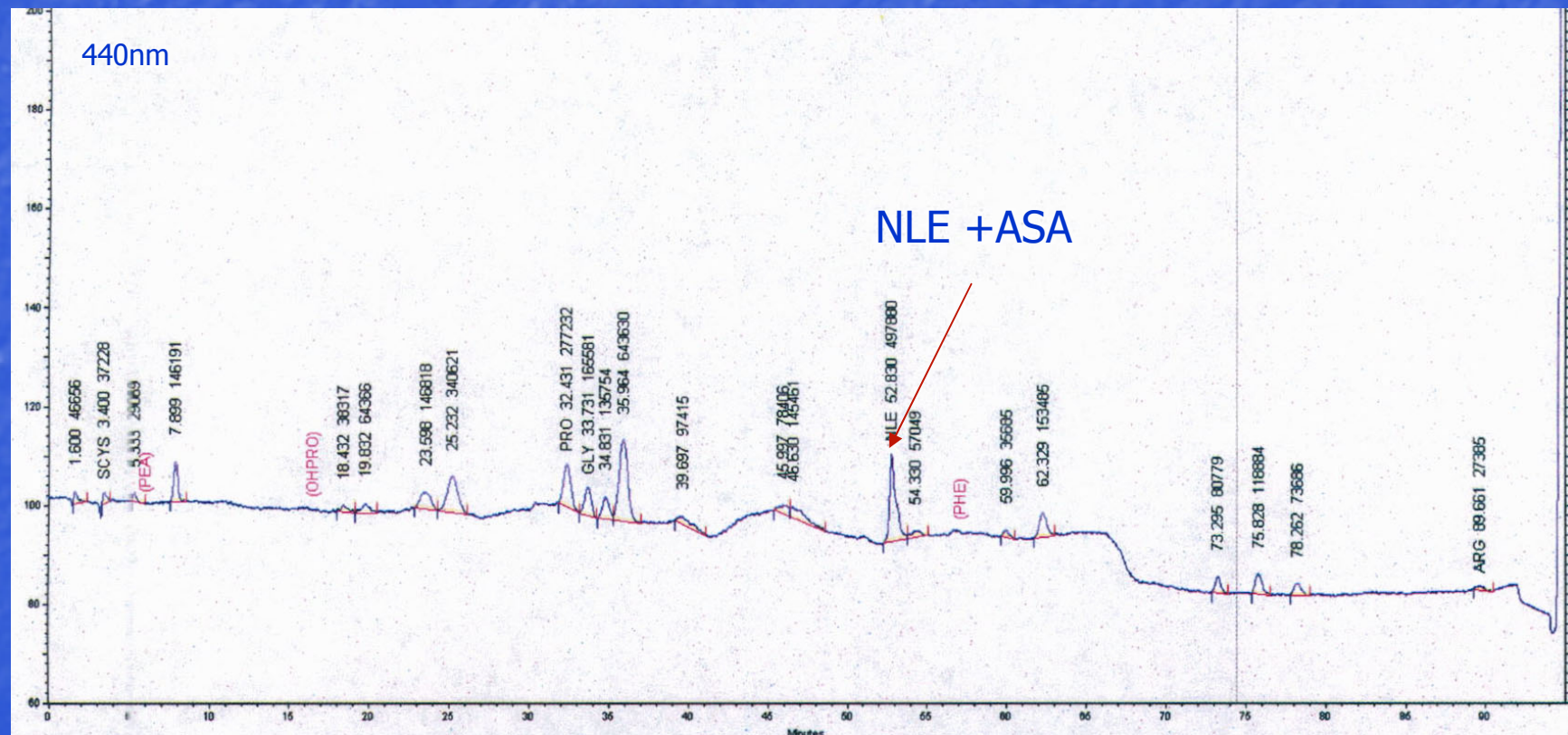
TLC on same sample using different ninhydrin stains



Plasma from patient with ASA



Same patient different plasma



Points to Remember:

- Look at Urine and Plasma samples
- TLC can aid identification
- Sulphocysteine is very hard to detect,
 - measure the uric acid and total homocysteine
 - look at amino acids on several samples, including CSF.
- Urine Organic acids should always be analysed
- Internal standard areas should be similar
- Utilise the "Branch Chain" program

Thank You

