

# Complications in reporting

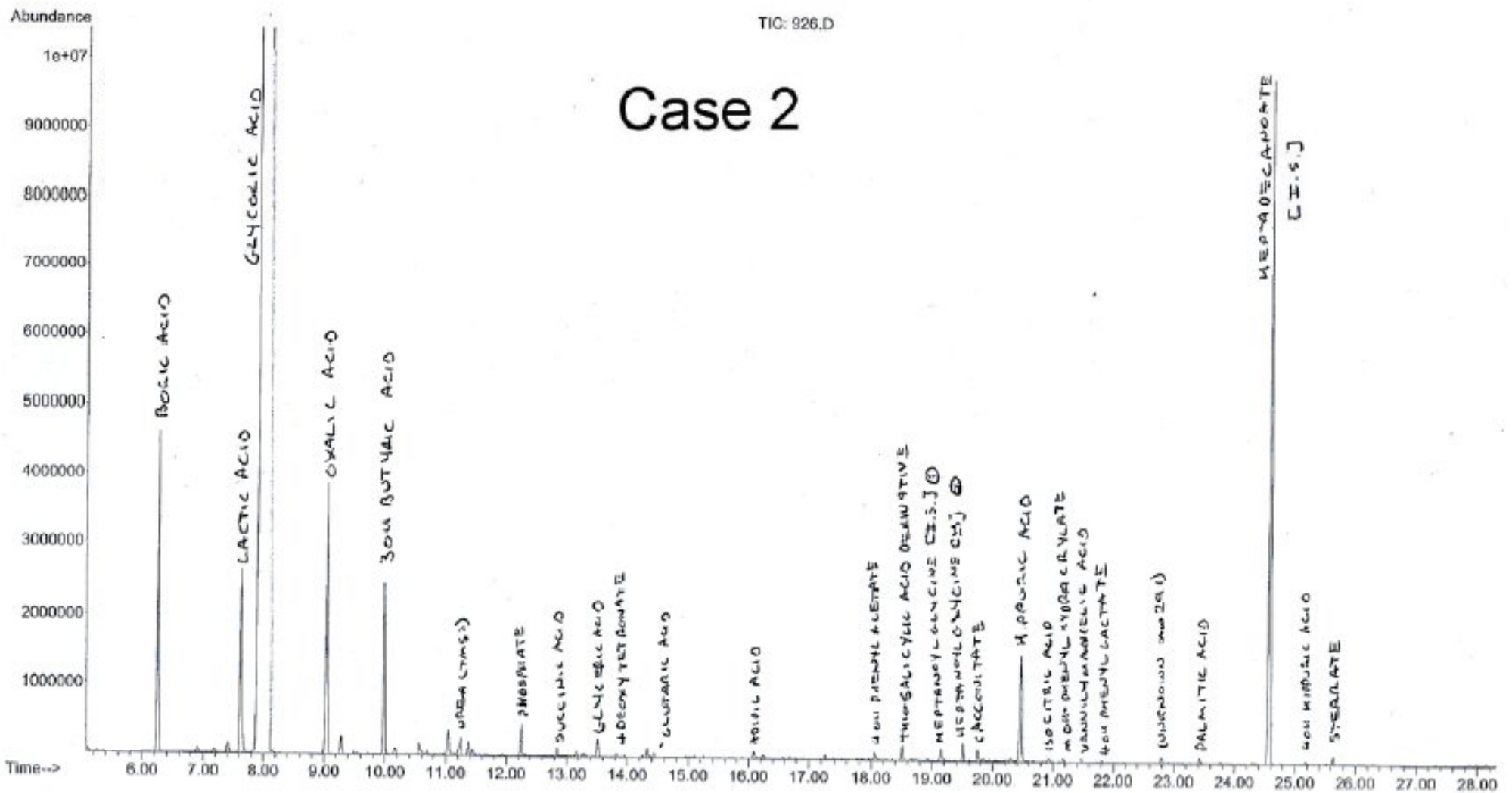
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# Outline of problem

- ERNDIM places high value on education, in addition to monitoring and improving quality
- A problem for Biochemical Genetics laboratories is deciding when and how to report information that may not directly relate to an IEM
- We feel that its important to recognise and report clinically significant information
- Good example; drugs

# Case 2



Clinical Details: Three year old boy, found at home acidotic and comatose

## *Patient 03.1, Sheffield DPT circulation 2006*

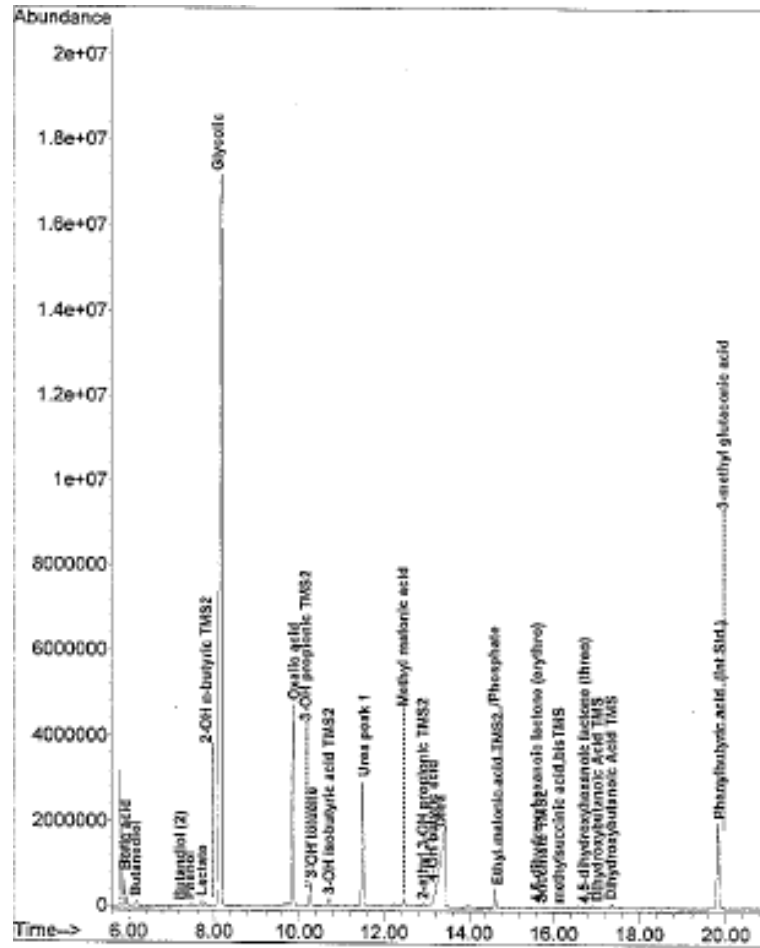
*Male patient, aged 28 years who presented with metabolic acidosis nausea and vomiting*

**The sample had been obtained from a patient who had ingested ethylene glycol**

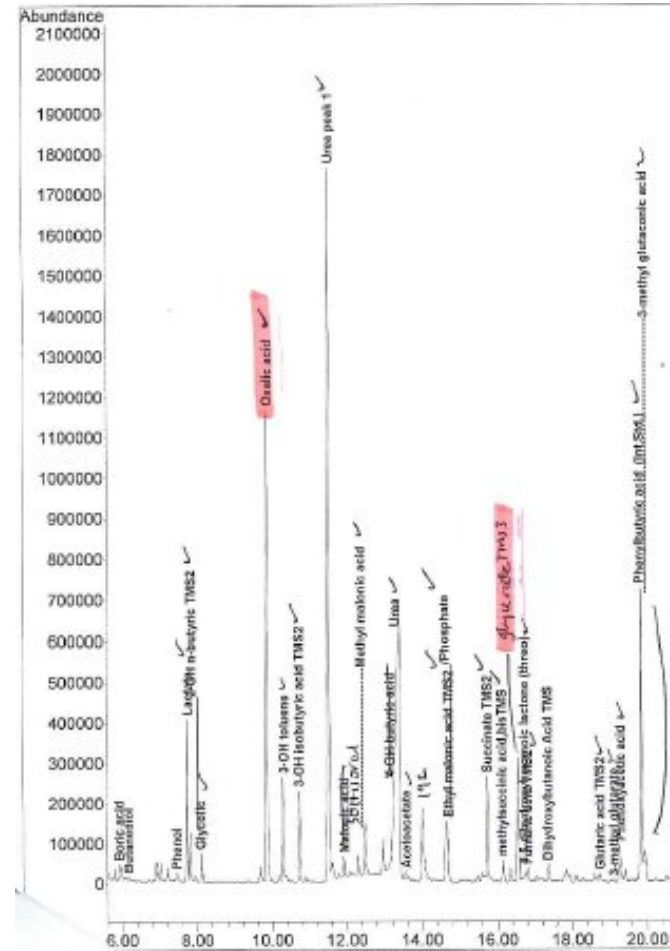
Results were returned from all 23 participants

- All 23 noted an increased excretion of glycolate
- 17 commented on increased oxalate
- 14 commented on increased lactate and hydroxy butyrate
- 22 indicated that these findings were consistent with ethylene glycol intoxication that would need urgent action.
  
- 14 indicated the need for further toxicological investigations to quantitate ethylene glycol in plasma to facilitate treatment
- 6 suggested measuring plasma calcium
  
- 14 highlighted the role of ethanol infusion for treatment
- 11 referred to the newer alcohol dehydrogenase inhibitor, *4-methylpyrazole*

## Hyperoxaluria type 1

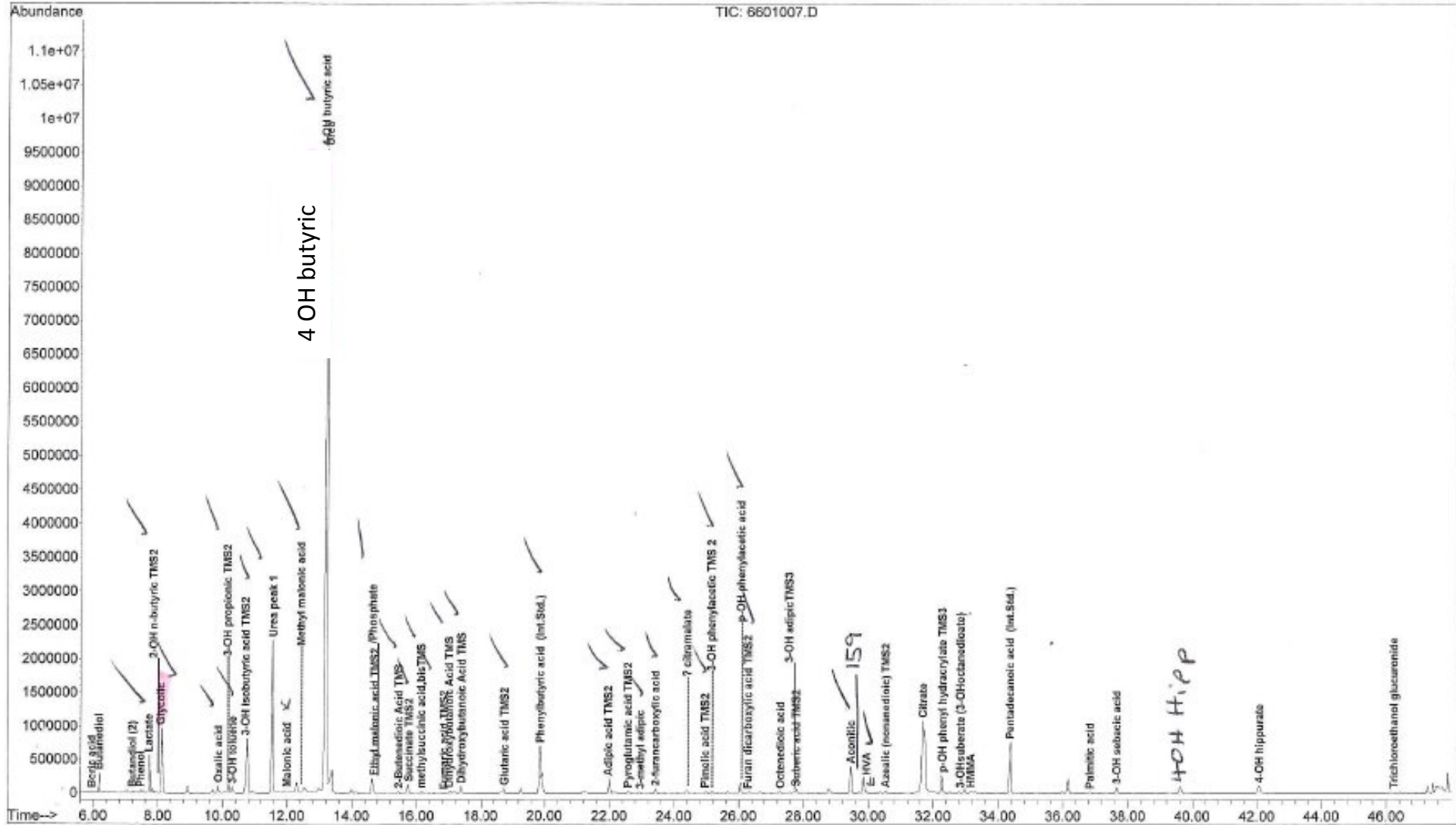


## Hyperoxaluria type 2



# Case 5

16 yr old male, unexplained loss of consciousness, bradycardia and reduced respiratory rate



## *Patient 139, Qualitative Organic Acid circulation 2006*

*Male patient, aged 16years who presented with unexplained loss of consciousness, bradycardia and reduced respiratory rate*

**The sample had been obtained from a patient who had taken the recreational drug 'GHB'**

Results were returned from all 60 of 65 participants

- 55 noted an increased excretion of 4-hydroxybutyrate (5 failed to identify this peak)
- 33 noted the presence of the metabolite 3,4-dihydroxybutyrate
- 10 noted the virtual absence of other metabolites, the 4,5 dihydroxyheptanoate lactones normally present in urine from patients with succinic semialdehyde dehydrogenase def.
  
- 45 indicated that their findings were consistent with 'recreational substance abuse'
- 30 suggested that this could be confirmed by repeat testing
  
- 29 concluded that this was from a patient with SSDH def and recommended enzyme assay and mutation testing