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A possible case of glycerokinase deficiency

HOSP #		WARD	Ward B2
CONSULTANT	George van der Watt	DOB/AGE	3 month

Abnormal Result

Glycerol which is significantly raised on urine organic acid analysis.

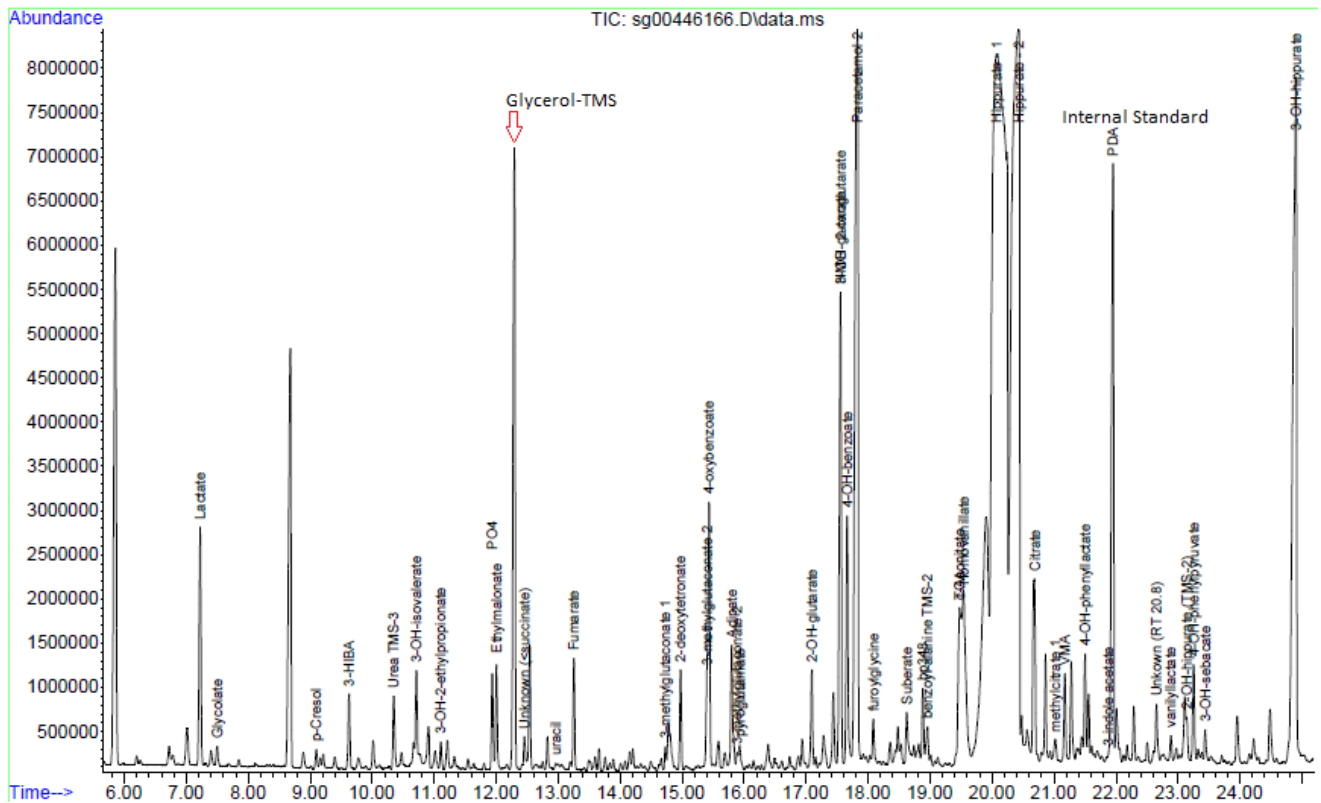


Figure 1 – Chromatogram. The high levels of Glycerol (with TMS – trimethyl silyl derivative) which is >0.5 the peak height of the internal standard (PCA – pentadecanoic acid).

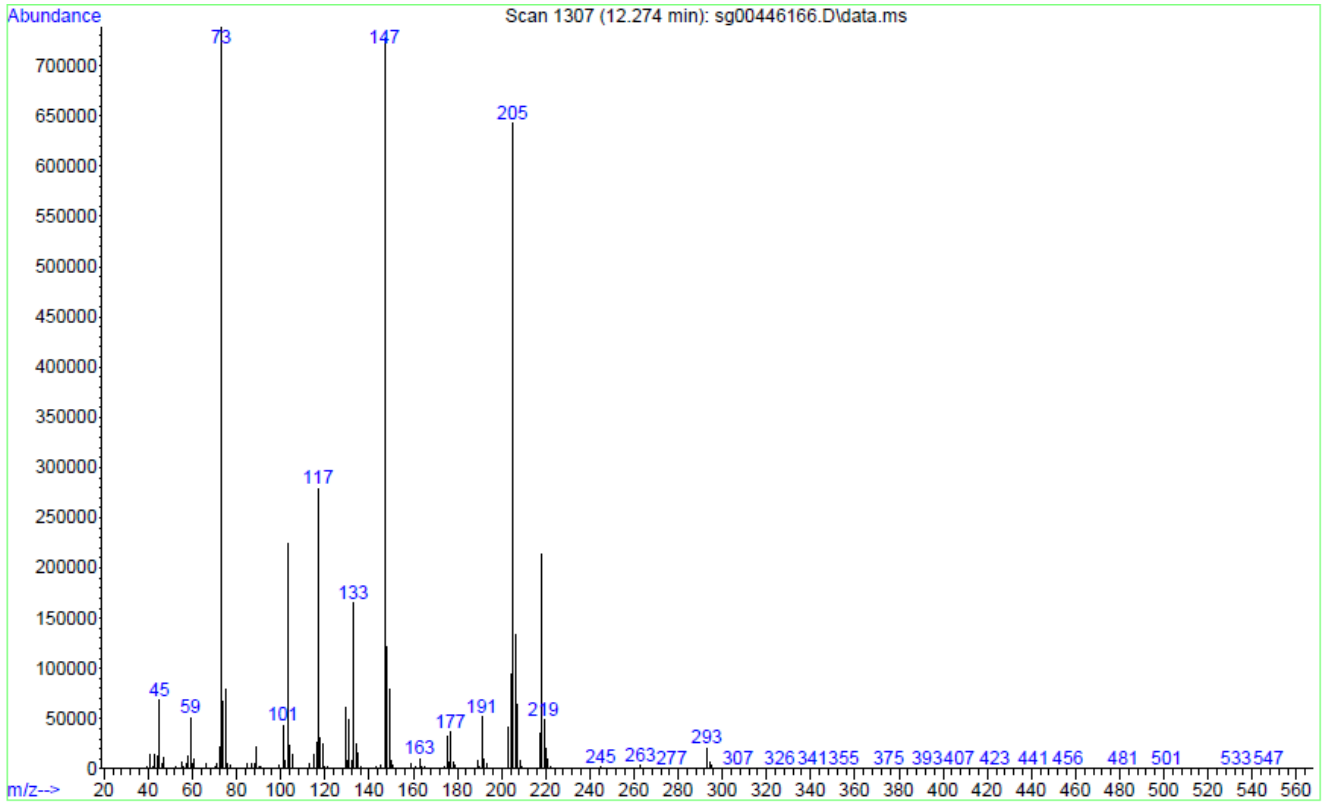


Figure 2 – Mass spectrum of the peak as indicated by Glycerol TMS above in Figure 1.

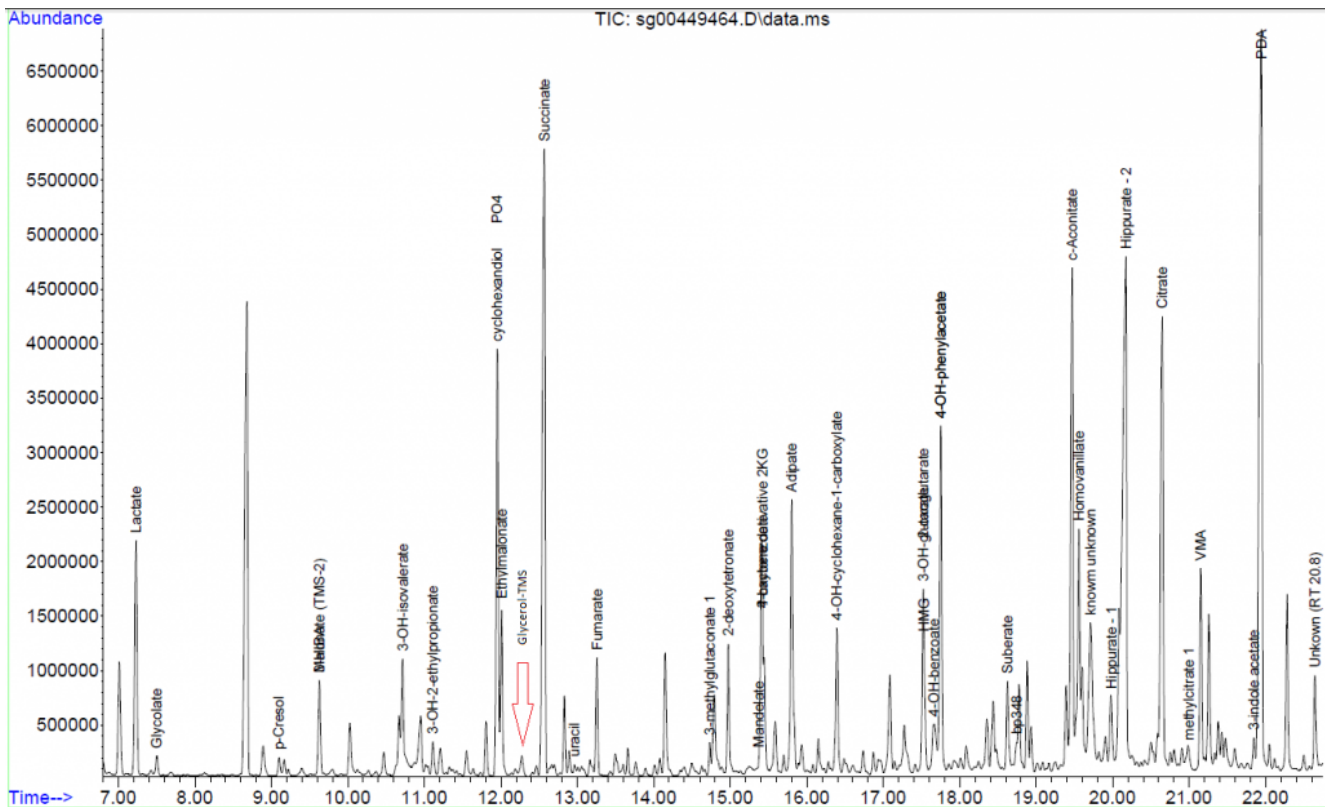


Figure 3 – Follow up gas chromatogram without KY-jelly

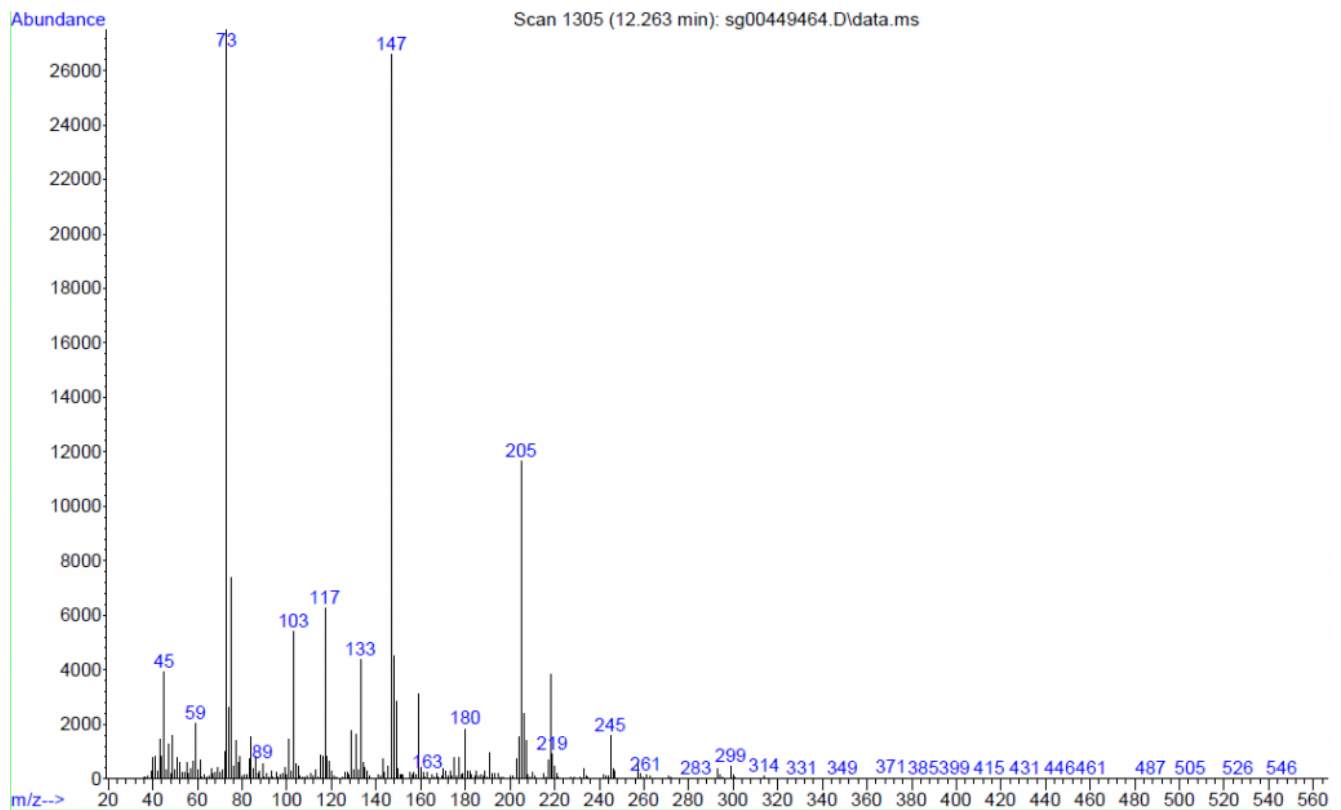


Figure 4 – Mass spectrum in the peak marked as “Glycerol-TMS” from figure 3.

Presenting Complaint

Patient is a 3 month old male with signs and symptoms of sepsis.

History

Patient presented with significant failure to thrive.

Laboratory Investigations

Triglycerides : 4.47 mmol/L

Other Investigations

Faecal elastase 81 ug/g stool

Reference range (adults and children > 1 month):

- > 200 ug elastase/g stool: Normal exocrine pancreatic function
- 100-200 ug elastase/g stool: Moderate/mild pancreatic insufficiency
- < 100 ug elastase/g stool: Severe exocrine pancreatic insufficiency

These ranges apply to formed stool samples. Watery stool samples may yield spuriously low elastase results due to dilution, and a formed stool sample should be sent for re-analysis.

Final Diagnosis

Glycerol contamination of the skin – as excluded by the repeat analysis.

Take Home Message

- Glycerol (glycerine) is a common contaminant of urine organic acids due to being present in various skin products / creams. Contamination can be eliminated by thorough cleaning of the perineum with normal saline or doing an “in-out” catheterization procedure for urine collection in neonates. Interestingly glycerol is also one of the main ingredients in KY jelly, a common lubricant use for catheterization.
- High glycerol in serum will present with a falsely high triglyceride level on most routine chemistry analysers due to the inherent enzymatic conversion of triglycerides to glycerol before further steps to measurement.

Triglyceride determination

- Oxidizing enzyme
 - Glycerol 3-phosphate oxidase (GPO)
 - FAD cofactor
- Conversion of triglycerides
- Oxidize glycerol 3-phosphate

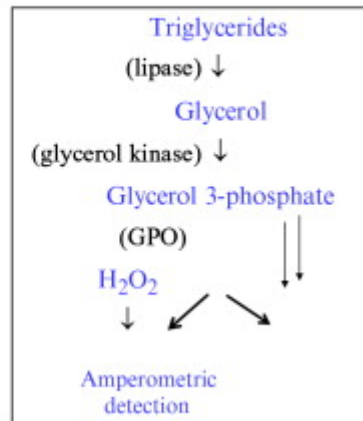


Figure 3 – Explanation of triglyceride determination by amperometric detection.

- Sepsis is more common than inherited metabolic diseases and so is pre-analytical caveats such as glycerol contamination of the perineal skin.

Hyperaldosteronism with Hyperreninaemia in a 15 year old

A case of hyperreninemia and hyperaldosteronism in a 15 year old. The cause will surprise you.