

A Case of Neurodevelopmental Delay

HOSP #		WARD	Neurodevelopmental clinic – Inkosi Albert Luthuli Hospital
CONSULTANT	Prof. George van der Watt	DOB/AGE	2y male

Abnormal Result

Urine organic acid analysis was performed upon which a big peak was seen, representative of phenylpyruvate.

Presenting Complaint

The patient was a 2 year old male evaluated at a neurology clinic for neurodevelopmental delay.

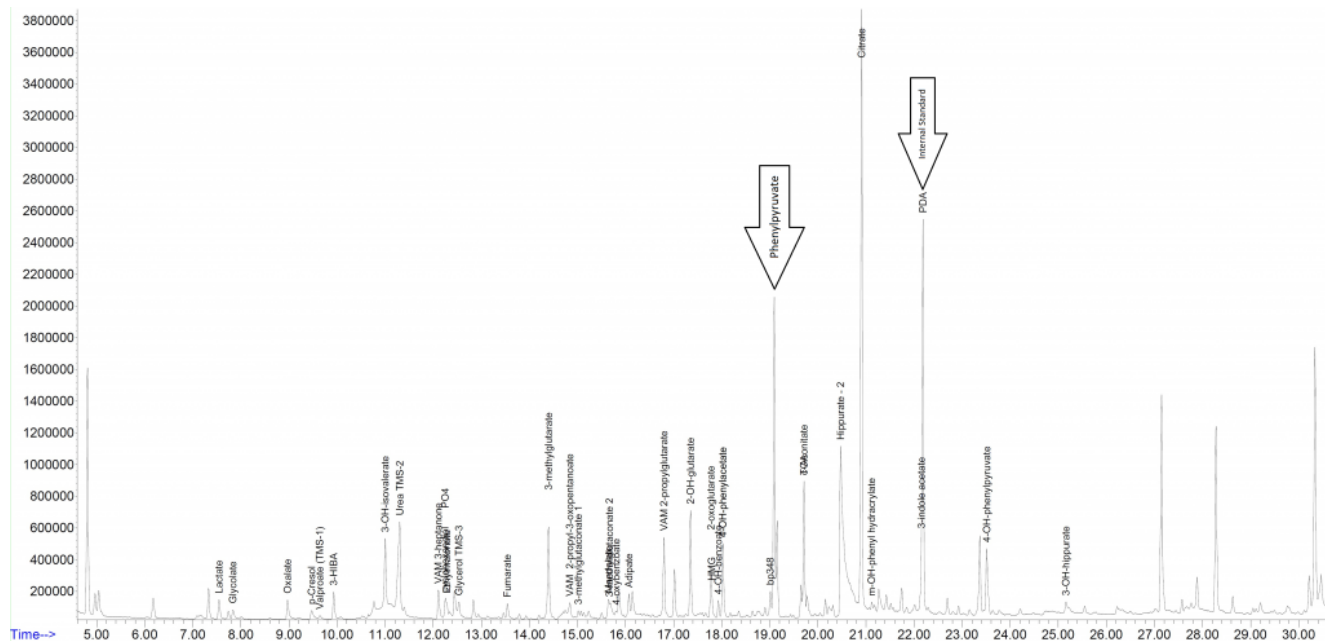
History

The patient's brother died at 3 or 4 years of age with similar neurodevelopmental delay.

Examination

Unfortunately this information was unavailable. The clinician I got hold of at Inkosi Albert Luthuli hasn't seen the patient himself.

Laboratory Investigations



Other Investigations

Final Diagnosis

The diagnosis is also supported by a plasma phenylalanine of 672 $\mu\text{mol/L}$ (ref < 67).

Take Home Messages

Phenylalanine levels $>600 \text{ umol/L}$ in serum is highly indicative of phenylketonuria

Prof. George van der Watt

Biopterin cycling defects usually cause levels >125 $\mu\text{mol/L}$.

This deficiency is 4-monooxygenase deficiency.

Management of PKU is with a phenylalanine restricted diet.