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

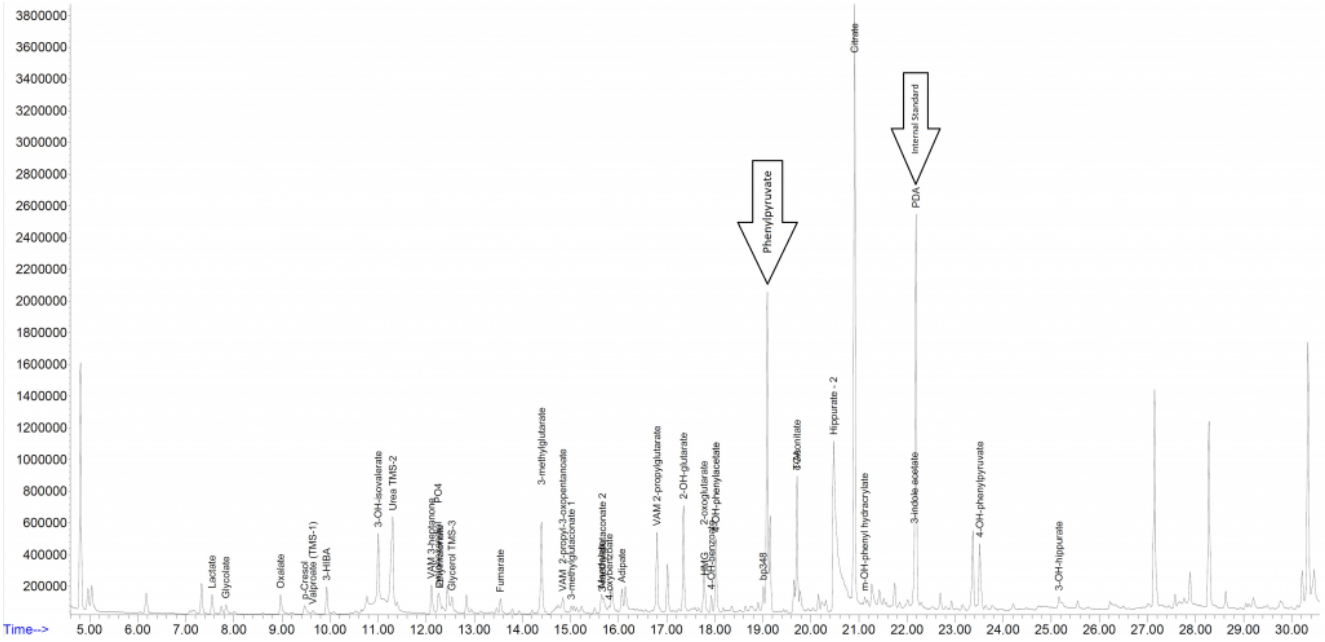

Fig 1 – Urine organic acid screening by GCMS demonstrates elevations of the phenylketones: phenylpyruvate and 4-OH phenylpuyruvate. These findings are indicative of a diagnosis of phenylketonuria due to autosomal recessive deficiency of phenylalanine hydroxylase.

Other Investigations

The urine amino acid analysis yielded a significantly raised phenylalanine: 672 umol/L (ref <67)

Final Diagnosis

This is a case of phenylketonuria

The diagnosis is also supported by a plasma phenylalanine of 672 umol/L (ref < 67).

Take Home Messages

Build-up of phenylalanine gets metabolised to phenylpyruvate (which is seen in urine at high levels).

> *Phenylalanine levels >600 umol/L in serum is highly indicative of phenylketonuria*

Prof. George van der Watt

Biopterin cycling defects usually cause levels >125 umol/L.

This deficiency is 4-monooxygenase deficiency.

Management of PKU is with a phenylalanine restricted diet.