Hypoglycemic seizures

HOSP #	MRN90378429	WARD	Endocrinology Ward
CONSULTANT	Jody Rusch	DOB/AGE	14 y girl

Abnormal Result

Fingerprick glucose 2.9 mmol/L

Presenting Complaint

Hypoglycemic seizure

History

The patient is a known type 1 diabetic patient who presented to the Internal Medicine Paediatric specialist OPD during two occasions of hypoglycemic seizures before.

The pateint had, according to the mothed, no post-ictal state.

She was admitted to the Endocrinology ward for a fast provocation test. At two hours, the glucose measured 2.9mM on point-of-care glucometer – glucose and other parameters on laboratory values however is illustrated below.

2 weeks after this presentation patient presented again with hypoglycemic seizures – mother is a nurse – puts in drip after which the patient’s condition normalizes.

IGF-1 normal, Ketones raised (quantitative beta-hydroxybutyrate, Insulin: 6 nmol/L, glucose: 3.5mM, hGH: 1.9 ug/L.

Examination

On examination the patient had no signs and symptoms of hypoglycemia (during the provocative test). And after the hypoglycemic seizure there were no “post-ictal” symptoms identified.

Laboratory Investigations

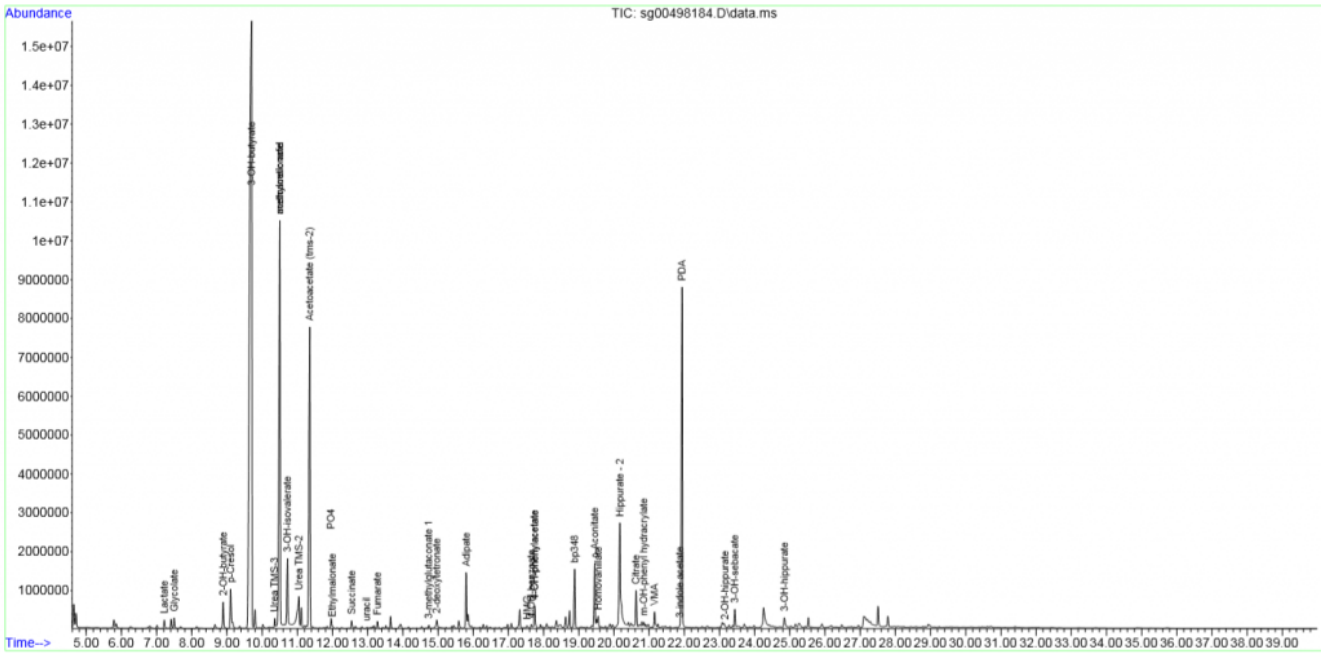
Glucose 3.5 mmol/L /L
Insulin: **6** nmol/L
Lactate 1.5 mmol/L (0.5 – 2.2)
Beta-hydroxybutyrate 1855 umol/L (20 – 270)
Ammonia 56 umol/L (11 – 35)
Cortisol 367 nmol/L
Cortisol reference intervals (when performed on a Roche Cobas analyzer):
Levels in adults: Morning (06:00-10:00) 133 – 537 nmol/L ; Afternoon (16:00-20:00) 68 – 327 nmol/L
Human growth hormone 1.9 ug/L

IGF-1 (Insulin-like growth factor I) @ 22/02/2021 09:30 : **366.0** ug/L (170.0 – 527.0)
Tanner stages Boys vs Girls:
Stage I 63 – 271 ug/L ; 71 – 394 ug/L
Stage II 114 – 411 ug/L ; 122 – 508 ug/L
Stage III 166 – 510 ug/L ; 164 – 545 ug/L
Stage IV 170 – 456 ug/L ; 174 – 480 ug/L
Stage V 161 – 384 ug/L ; 169 – 400 ug/L

Synacthen stimulation test:

Time on 22/02/2021	Cortisol (nmol/L)
14h00 (Baseline)	316
14h30	597
15h00	436

Other Investigations



Urine organic acid analysis profile: The 3 prominent peaks on the left are from left to right: B-hydroxybutyrate, Acetoacetate (with TMS derivative 1), Acetoacetate 2nd peak (with TMS derivative 2). TMS = trimethylsilyl derivative reagent, PDA = pentadecanoic acid (internal standard).

Final Diagnosis

Type 1 Diabetes with ketoacidosis and occasional episodes of hypoglycemia

Possible reasons for the hypoglycemia may be:

1. Ketogenic diet (fairly easily excludable I think).
2. Ketone utilisation disorder:
   - SCOT (succinyl-CoA:3-ketoacid CoA transferase) deficiency
   - Mitochondrial acetoacetyl-CoA thiolase (beta-ketothiolase) deficiency

Take Home Message

There are two predominant ketone utilisation disorders: SCOT deficiency and beta-ketothiolase deficiency. These disorders produce fairly continuous ketones, as they cannot be metabolised in the muscle and brain upon these deficiencies, which are autosomal recessive (as is most inherited metabolic diseases).

Giving the mother a urine dipstick home to measure urine at home mane before meals, midday just before meals and late afternoon or so before meals may be advised to assist with the diagnosis of one of the above disorders.

Urine organic acid analysis can sometimes pick up a marker to diagnose beta-ketothiolase deficiency:

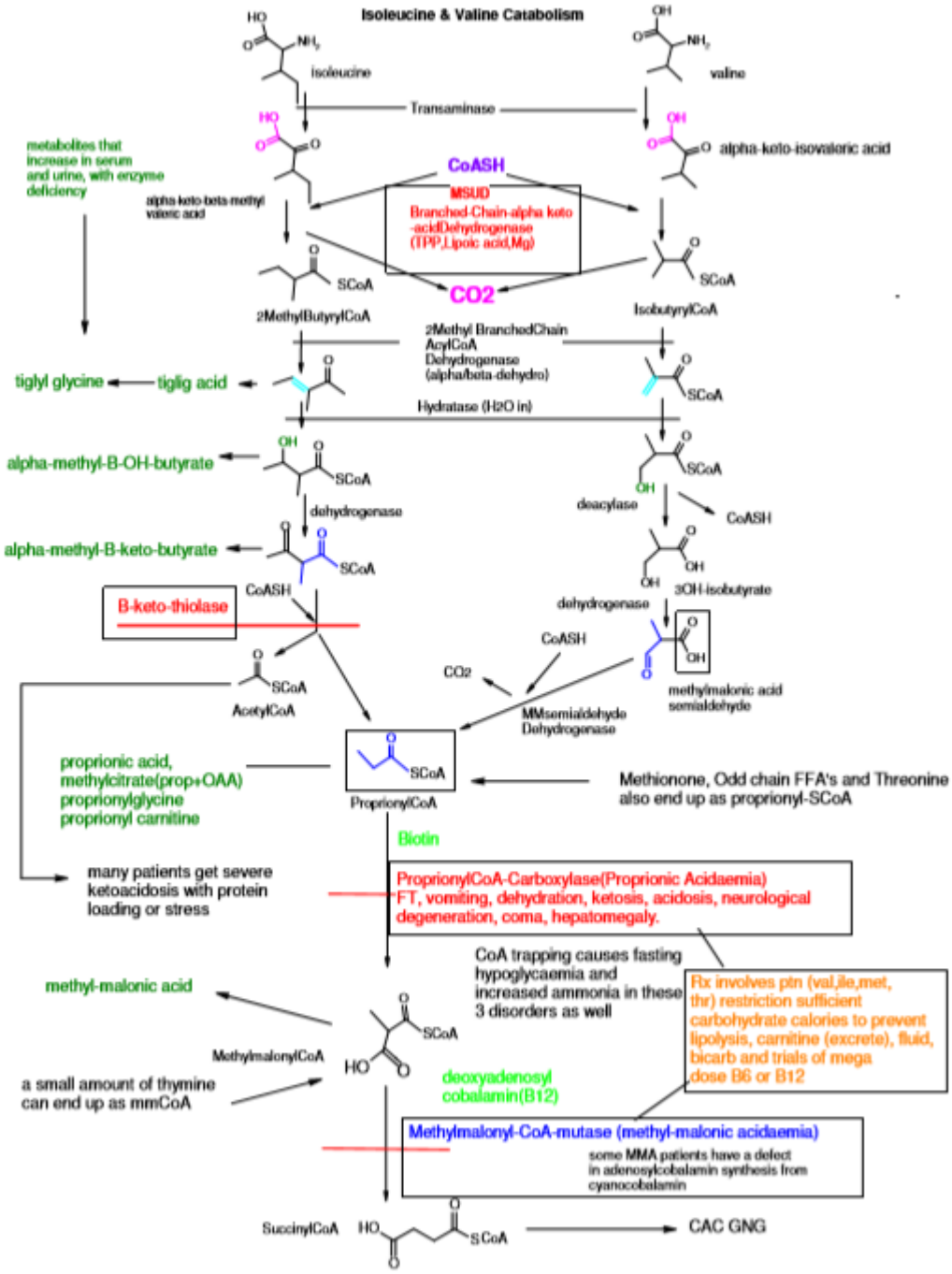


Figure 2 – The metabolism of Isoleucine and valine (credits to George van der Watt). In beta-ketothiolase deficiency, alpha-methyl-beta-keto-butyrate will accumulate, and can be detected on urine organic acid analysis by GC-MS.