A likely case of thyrotoxic periodic paralysis

Patient presented with a few isolated episodes of muscle weakness. This progressed from 2 weeks before, during the index episode, to become so severe that he couldn’t walk.

Conn’s syndrome with a focus on a unilateral adrenal gland

HOSP #	Mrs DW	**WARD**	Endocrine Department – CathLab – UCT private Hospital
CONSULTANT	Dr Jody Rusch	**DOB/AGE**	49y Female

Abnormal Result

49yr old female

Presenting Complaint

Medical complaint: Suspected Conn’s disease – right adrenal lesion/ irregular left adrenal gland

History

Past Medical History: Resistant Hypertension, primary hyperaldosteronism (confirmed previously with saline infusion

test), hypokalaemia, hypercholesterolaemia, newly diagnosed DM.

Family History: Hypertension – Mother.

Past Surgical History: TAH – 7 years ago.

Allergies: Nil known

Smoker

Meds: Amlodipine/Valsartan 10/320 daily, Doxazosin 8mg daily, Furosemide 40mg daily, Spironolactone 25mg daily, Carvedilol 25mg daily, Metformin 1g nocte, Simvastatin 20mg nocte, Zolpidem 10mg nocte.

Examination

Not available

Laboratory Investigations

	Time	Aldosterone Episode	Aldosterone pmol/L	Cortisol nmol/L	Selectivity Index Cortisol AV/PV	ACR	Lateralisation Index Dom A/C : nonDom A/C	Mean Aldo/Cort RAV	Aldo/Cort LAV
RAV 1	10:10	SA04663221	1310	659	0.2	2.0			
RAV 2	10:36	SA04663224	1490	681	0.2	2.2			
RAV 3	10:36	SA04663229	INS	712	0.2	#VALUE!			
RAV 4	10:39	SA04663232	771	340	0.1	2.3			
RAV 5	10:49	SA04663235	1470	692	0.2	2.1			
Mean RAV			1260.25	616.8	0.2			2.0	
LAV 1	10:01	SA04663256	2160	10790	3.0	0.2	0.1		0.2
LAV 2	10:02	SA04663250	3210	14540	4.0	0.2	0.1		0.2
LAV 3	10:03	SA04663242	5260	2621	0.7	2.0	1.0		2.0
LAV 4	10:59	SA04663238	2760	11870	3.3	0.2	0.1		0.2
LAV 5	10:03	SA04663246	3590	10770	3.0	0.3	0.2		0.3
Mean LAV			3396	10118.2	2.8				0.3
PIVC 1	11:00	SA04663213	2540	3609					
PFEM 1	9:43	SA04663217	803	301					
Arm	10:12	SA04663208	1330	724					

Key:

RAV	Right Adrenal Vein
LAV	Left Adrenal Vein
PIVC	Peripheral Inferior Vena Cava
PFEM	Peripheral Femoral Vein
UTC	Unable to calculate
*	Not assayed in dilution
AV/PV	Adrenal Vein to Peripheral Vein Ratio
ACR	Adrenal to Cortisol Ratio

Peripheral	
Aldosterone	2540
Cortisol	3609

Other Investigations

Not available for this patient.

Ideally one would need a CT with contrast beforehand to adequately visualize the positions of the adrenal veins, as this may aid in the canulation, especially of the right adrenal vein.

One needs to diagnose hyperaldosteronism (by an appropriate salt loading test) before proceeding to bilateral adrenal vein sampling.

Final Diagnosis

Interpretation

Definition	Formula	Clinical significance
Selectivity index	PCC(side) / PCC (ivc)	>cutoff confirms canulation of adrenal vein >3 stimulated >2 unstimulated
Lateralization index	PAC/PCC (dom) : PAC/PCC (non-dom)	>cutoff confirms laterilization of hyperaldo secretion >4 stimulated >2 unstimulated
Contralateral suppression index	PAC/PCC (non-dom) : PAC/PCC (ivc)	<cutoff indicate ipsilateral suppression and suggest contralateral aldosterone overproduction.

Table 1 – Interpretation of bilateral adrenal vein sampling. PCC: plasma cortisol concentration, PAC: plasma aldosterone concentration, ivc: inferior vena cava or peripheral vein,

dom: dominant side, non-dom: non-dominant side.

Selectivity index

Right: 0.2 (mean)

Left: 2.8 (mean)

These two results indicate that the left adrenal has likely been canulated adequately, but the right vein inadequately.

Lateralization index

Unable to comment because of the inadequate canulation of the right adrenal vein. If determined, it would very likely provide a false result.

Contralateral suppression index

616.8 /1260.25 : 2540/3609

= 0.70

This falls in between some of the referenced cutoffs (<1 and <0.5)

All of the other samples also fall somewhere in this range. Biochemically, these results suggests inadequate right sided venous sampling (a commonly described problem)

Take Home Message

- Procedure is done in the Cath Lab
- The patient received continuous synacthen infusion
- Done under imaging with contrast used for the localisation of the adrenal gland and adrenal vein
- Sequential sampling technique used, generally > 20 mins infusion
- Multi-disciplinary: nurses, anaesthetist, radiographer,

intervention radiologists, students, chemical pathologists
- Difficulty with sampling right side for both patients
- Difficulty with interpreting results – most likely due to inadequate canulation of the right adrenal vein

Some important learning points

1. Adrenal vein sampling may be a valuable tool that is underutilised
2. Careful selection of patients essential – also patient should consent to surgical removal of the affected adrenal before this invasive procedure is initiated
3. Inter-disciplinary approach is necessary
4. Obtaining cosyntroponin (aka synacthen) can be difficult (Section 21), but recommended
5. Right adrenal access difficult: may require specific imaging. Recommended to start on the right or do simulataneous sampling
6. Adrenalectomy may be curative or help achieve better control of BP thus decrease associated morbidity and mortality in those with unilateral adenoma

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Amiodarone-induced hyperthyroidism

An interesting case of Amiodarone – induced hyperthyroidism

A case of amenorrhoea in a 17-year old female

A case of hyperprolactinemia with amenorrhoea

Laughing spells and precocious puberty in a child

A case of laughing spells with precocious puberty

A case of high HDL-cholesterol

HOSP #		**WARD**	GP Clinic
CONSULTANT	John Stanfliet / Jody Rusch	**DOB/AGE**	73 year Female

Abnormal Result

Abnormal lipid profile (see below)

Presenting Complaint

A 73 year old female was investigated with a full lipid profile after presenting with an increased total cholesterol

upon routine screening at her general practitioner.

History

The patient had an increased Total Cholesterol, but was otherwise not unwell. Medication history unfortunately not available.

Examination

Not available

Laboratory Investigations

Test	Result
Urea	7.2 mmol/L
Creatinine	105 umol/L
eGFR	46 ml/min/1.73m2
Fasting Lipid profile (lipemia index - turbidity- on sample was absent):	
Total Cholesterol	6.7 mmol/L
Triglyceride	0.6 mmol/L
HDL Cholesterol	> 4.7 mmol/L
Non-HDL Cholesterol (calculated)	< 2.0 mmol/L
LDL Cholesterol (calculated)	< 1.7 mmol/L
LDL Cholesterol (direct – measured)	1.3 mmol/L
Glucose Fasting	5.5 mmol/L

Table 1 – Full lipogram with other routine chemistry tests.

Other Investigations

To rule out the possibility of interferents, the following tests were performed.

Test	Value
Apo A1	4.24 g/L (424 mg/dL) (Ref. >140 mg/dL)
Apo B	0.52 g/L (52 mg/dL) (Ref. < 130 mg/dL)
Apo B : Apo A1 ratio (calculated)	0.12

Table 2 – ApoA1 and ApoB by immunoassay. ApoA1: the major lipoprotein in HDL particles. ApoB: the major lipoprotein in Non-HDL particles.

Final Diagnosis

Increased HDL which may likely be an APOC3 deficiency.

Take Home Message

Although not present in this case, elevated apolipoprotein B (ApoB) confers increased risk of atherosclerotic cardiovascular disease, even in a context of acceptable LDL cholesterol concentrations. Extremely low values of ApoB (<48 mg/dL) are usually related to malabsorption of food lipids and can lead to polyneuropathy. Reduced apolipoprotein A1 (ApoA1) confers an increased risk of coronary artery disease. Extremely low ApoA1 (<20 mg/dL) is suggestive of liver disease or a genetic disorder. Elevated ApoB:ApoA1 ratio confers increased risk of atherosclerotic cardiovascular disease, independently of LDL and HDL cholesterol concentrations.

If the inverse of the above is true, then this lady is likely destined to live forever, but that’s the whole conundrum in lipid metabolism – the inverse of one’s theories does not always hold true under randomized controlled studies, and due to the difficulty of finding a proper control group. It was however previously demonstrated that patients with ApoC3 deficiency (if this is the cause in this case) increases

longevity.

> *APOC3 has been established as an inhibitor for lipoprotein lipase, a gene that hydrolyzes triglycerides to generate free fatty acids before their uptake by muscle and adipose tissue (reviewed in Jong et al). Mice with a high-level expression of human APOC3 on a background of Ldlr deficiency proved to be an excellent model for familial combined hyperlipidemia, because they are disturbed in the breakdown of triglycerides. In contrast, mice lacking Apoc3 show increased activity of LPL, which causes hypotriglyceridemia and protection from postprandial hypertriglyceridemia. From these mice studies, it became clear that a deficiency of APOC3 could cause a healthier lipoprotein profile, which is associated with protection from cardiovascular diseases. However, in the absence of APOC3-deficient subjects, this hypothesis was difficult to test directly.*
>
> *Dodacki, A., Wortman, M., Saubaméa, B. et al. Expression and function of Abcg4 in the mouse blood-brain barrier: role in restricting the brain entry of amyloid-β peptide. Sci Rep* ***7,*** *13393 (2017). https://doi.org/10.1038/s41598-017-13750-0*

Glucagon Stimulation C-peptide testing

HOSP #		**WARD**	Endocrinology ward
CONSULTANT	Dr. Heleen Vreede	**DOB/AGE**	22 y Male

Abnormal Result

Patient presented with Diabetic Ketoacidosis and a glucose value of 27.4 mmol/L.

Presenting Complaint

Signs and symptoms typical of Diabetic Ketoacidosis

History

Patient was diagnosed with diabetes 7 years ago after presenting with diabetic ketoacidosis. Upon diagnosis he was given insulin in the hospital. Upon discharge he was given Metformin and Glimeperide (oral hypoglycemic medication – reason for oral agents unknown – likely because of his young age?). Defaulted Rx completely. Presented with DKA again. Restarted about 2 y ago on insulin.

The differential diagnosis at the current presentation is thus one of:

1. Ketosis prone diabetes
2. LADA (Latent auto-immune diabetes of the adult)
3. Type1 – went into honeymoon phase after diagnosis and now relapsed

To differentiate – the clinicians prompted to do antibodies, insulin levels and a glucagon stimulation c-peptide dynamic test.

Examination

N/A

Laboratory Investigations

Date	05/02/2021	02/02/2021	28/08/2018	25/01/2018	05/06/2017	03/03/2017	03/03/2017	24/02/2017	16/09/2016	14/04/2015
Na		134 L					137.000			133 L
K		4,6					4.890		4,5	UOLD2
Cl		93 L								
Urea		13,4 H					5.000			1,9 L
Creat		91					69.000		66	34 L
Glu Random				27,4				21.860		
HbA1c (NGSP)		12,7	>14	13,7	>14			12,8	13,7	
Total chol	5,04						ბ- 4,98			
Triglyceride	1,74						1,25			
HDL chol	1,35						1,16			
LDL chol (calc)	2,89						3,25			
Total chol									8,99	
U creat	4,1						1,9			
U albumin	32.70						<3			
U alb : creat	8.0 H						UTC			
Test referred						Anti-IA2 Antibody Positive; Anti-GAD antibody Positive				

Other Investigations

A glucagon-stimulated C-peptide level was drawn.

0 min	1.5 ug/L	0.5 nmol/L
1 min	2.0 ug/L	0.67 nmol/L
2 min	1.9 ug/L	0.63 nmol/L
3 min	1.9 ug/L	0.63 nmol/L

Final Diagnosis

LADA – latent autoimmune diabetes of the adult

Take Home Message

Serum c-peptide has traditionally been thought to be an inconvenient method as immediate lab analysis is required before degradation (when collected in serum gel or plain sample tubes). This is because c-peptide is a small linear peptide, which is susceptible to enzyme proteolytic cleavage.

Gel tubes are therefore traditionally required to be transported to the lab on ice, promptly centrifuged and separated, then stored in frozen conditions unless lab analysis is possible at that center.

However, c-peptide sample collection for c-peptide determination in whole blood in EDTA prepared tubes is stable at room temperature for up to 24 h. Venous blood c-peptide levels can be measured in the random, fasting, or stimulated scenarios. Random samples are taken at any time during the day without consideration of recent food intake, whereas fasting samples are taken after an 8- to 10-h fast.

Stimulation methods include using

- glucagon
- intravenous/oral glucose
- tolbutamide
- sulfonylurea
- glucose-like peptide 1
- amino acids
- a mixedmeal

In this case a glucagon stimulation yielded sufficient results to assist the clinicians in making the diagnosis, indeed a case of atypical diabetes presentation.

An uncommon cause of unconjugated

hyperbilirubinemia

HOSP #		WARD	Red Cross Endocrinology
CONSULTANT	Dr Jody Rusch	DOB/AGE	27 day female

Abnormal Result

TSH > 100

Free T4: 0.5 pmol/L

Presenting Complaint

Patient was brought to the ER being lethargic.

History

Term Neonate; Had a history of profound jaundice after birth, with unconjugated hyperbilirubinemia.

The patient’s mother lives in Athlone, gave birth at Carl Bremer hospital where a cord blood TSH was done, but results not available at the time.

Examination

No overt abnormalities on examination was found, except the single sign of jaundice.

No defects at the base of the tongue was observed.

No abnormalities in the neck was observed.

Laboratory Investigations

TSH > 100

Free T4: 0.5 pmol/L

Other Investigations

The patient had an ultrasound of the abdomen done (since it was the first occurence of hyperbilirubinemia, and in fact is termed pathological jaundice).

Cord blood TSH was retrospectively reviewed as being 178 uIU/ml.

Final Diagnosis

Congenital hypothyroidism

Take Home Message

Congenital hypothyroidism (**CH**) is thyroid hormone deficiency present at birth. If untreated for several months after birth, severe congenital hypothyroidism can lead to growth failure and permanent intellectual disability. Infants born with congenital hypothyroidism may show no symptoms, or may display mild symptoms that often go unrecognized as a problem. Significant deficiency may cause lethargy, hypotonia, hoarse cry, infrequent bowel movements, significant jaundice, and hypothermia.

Causes of congenital hypothyroidism include

- iodine deficiency (most common cause)
- developmental defect in the thyroid gland, either due to a genetic defect or a biochemical defect in thyroxine production
- pituitary defects – congenital hypopituitarism (present at birth) may be the result of complications around delivery, or may be the result of insufficient development (hypoplasia) of the gland, sometimes in the

context of specific genetic abnormalities.

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

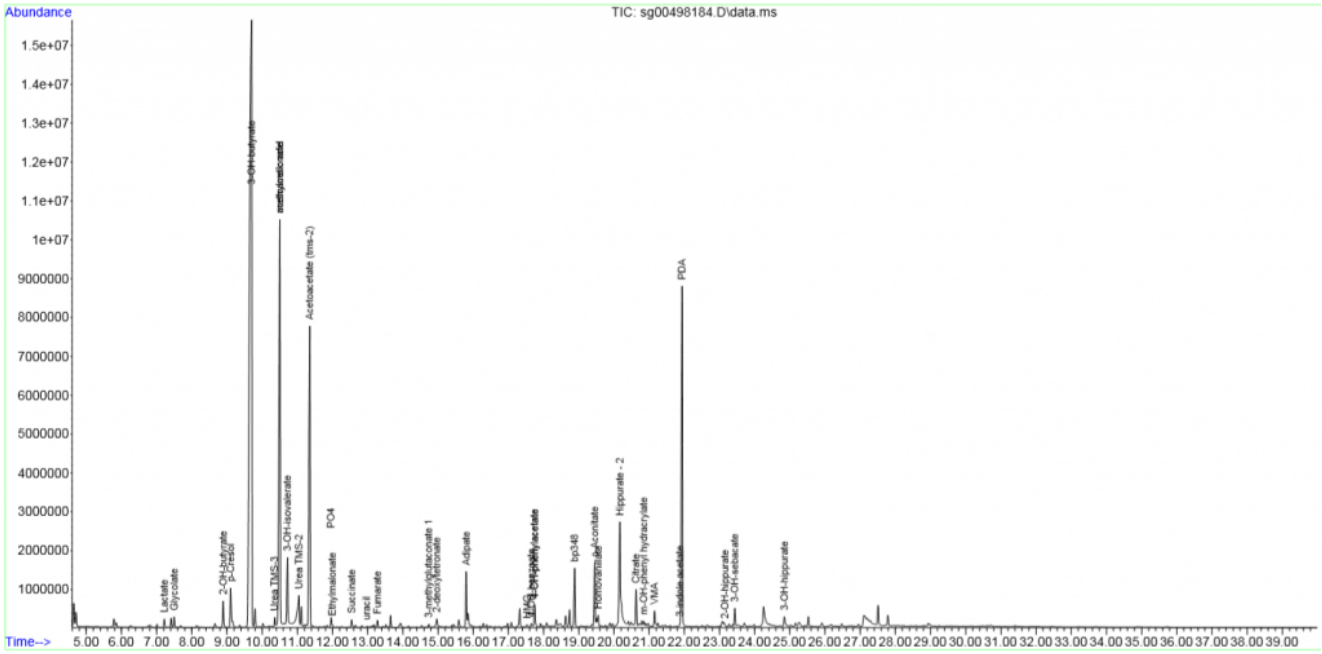
IGF-1 (Insulin-like growth factor I) @ 22/02/2021 09:30 : **366.0** ug/L (170.0 – 527.0)
Tanner stages Boys vs Girls:
Stage I 63 – 271 ug/L ; 71 – 394 ug/L
Stage II 114 – 411 ug/L ; 122 – 508 ug/L
Stage III 166 – 510 ug/L ; 164 – 545 ug/L
Stage IV 170 – 456 ug/L ; 174 – 480 ug/L
Stage V 161 – 384 ug/L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

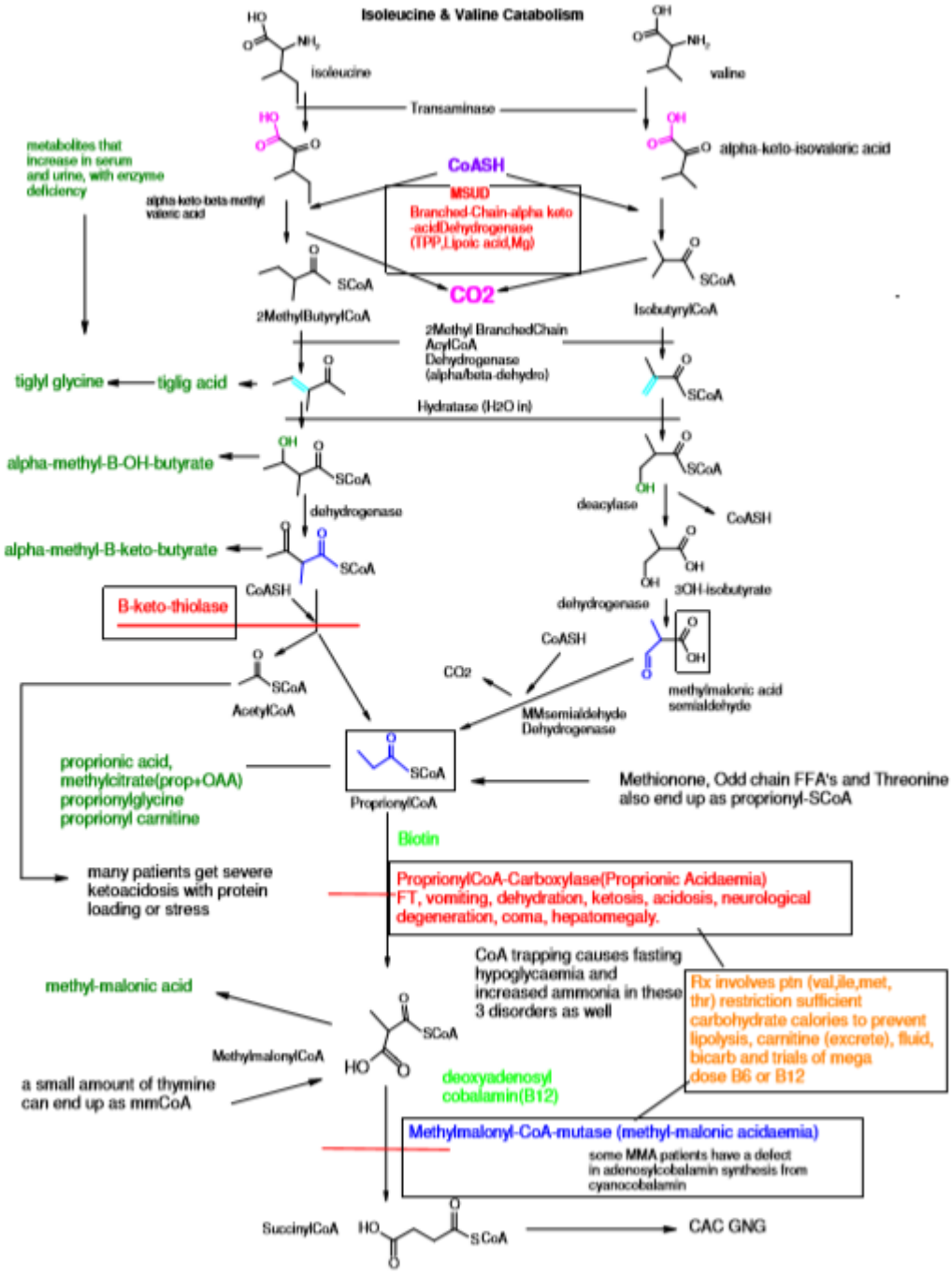


Figure 2 – The metabolism of Isoleucine and valine (credits to George van der Watt). In beta-ketothiolase deficiency, alpha-methyl-beta-keto-butyrate will accumulate, and can be detected on urine organic acid analysis by GC-MS.

A classic case of Cushing Disease

A classic case of Cushings Disease

An interesting cause of hyponatremia

HOSP #		**WARD**	Red Cross Hospital Oncology ward
CONSULTANT	Dr Amith Ramcharan / Dr Jody Rusch	**DOB/AGE**	11y Female

Abnormal Result

Persistent hyponatremia

2018 supracellar JPA (Astrocytoma)

Seizures – phenobarb.

Chemo @ 8 y of age.

Vincristin and Carboplatin administration

Craniospinal radiation – leptospinal

Presenting Complaint

Seizures – controlled with Phenobarbital

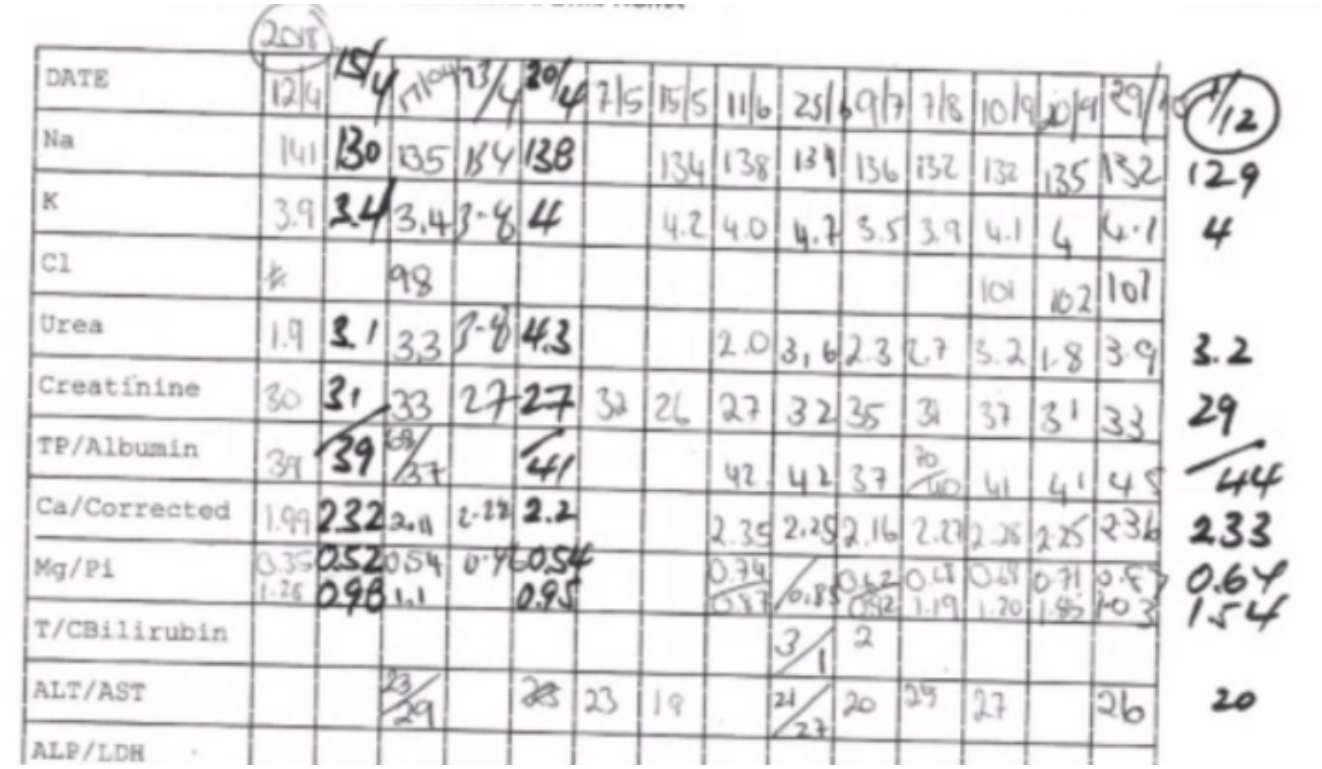
History

This is an 11 year old patient with a suprasellar JPA (Juvenile Pilocytic Astrocytoma). The tumour was diagnosed at 8y of age, upon which chemotherapy with Vincristine and Carboplatin was initiated. The pituitary was close to the area of radiation therapy as well.

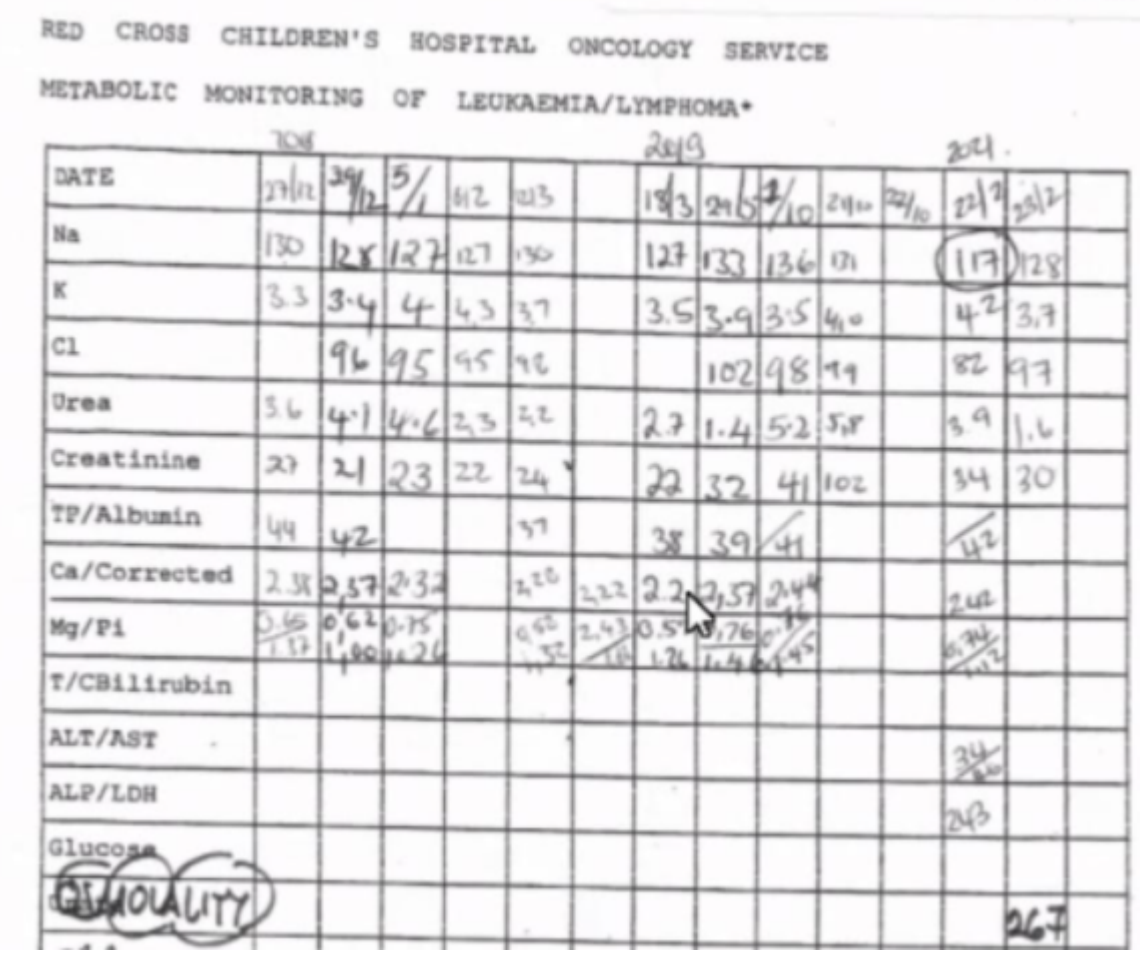
Examination

The patient’s hydration status was normal and there was no cerebral edema.

Laboratory Investigations



2018 – Electrolytes relatively stable



RED CROSS CHILDREN'S HOSPITAL ONCOLOGY SERVICE

METABOLIC MONITORING OF LEUKAEMIA/LYMPHOMA*

DATE	27/12	3/1/19	5/1	612	1013		18/3	29/5	1/10	29/10	24/10	22/12/20	25/12	
Na	130	128	127	127	130		127	133	136	131		117	128	
K	3.3	3.4	4	4.3	3.7		3.5	3.9	3.5	4.0		4.2	3.7	
Cl		96	95	95	98			102	98	99		82	97	
Urea	3.6	4.1	4.6	2.3	2.2		2.7	1.4	5.2	5.8		3.9	1.6	
Creatinine	27	21	23	22	24		22	32	41	102		34	30	
TP/Albumin	44	42			37		38	39	41			42		
Ca/Corrected	2.35	2.37	2.32		2.26	2.22	2.2	2.57	2.44			2.62		
Mg/Pi	0.65 / 1.17	0.62 / 1.80	0.75 / 1.26		0.92 / 1.32	2.43 / 1.0	0.5 / 1.26	0.76 / 1.46	0.9 / 1.45			0.34 / 1.12		
T/CBilirubin														
ALT/AST												34 / 30		
ALP/LDH												243		
Glucose														
[illegible] QUALITY													267	

2018-2019 – Hyponatremia and hypomagnesemia developing
The patient was found to have hypothyroidism and started on T4 replacement 50ug mane.

Other Investigations

Urine electrolytes on 23/02/2021:

- Na 54 mM
- K 31.3 mM
- Cl 110 mM
- Osmol 554 mOsmol
- Fractional reabsorption of phosphate: 85%

Final Diagnosis

Unknown – but likely indicates a tubular loss of sodium due to the chemotherapeutic agent(s).

Take Home Message

Chemotherapeutic agents does cause tubulopathy.

TMP/GFR is likely a better indicator of renal phosphate handling than only fractional reabsorption of phosphate. This can be calculated mathematically or read from a nomogram.

Falsely decreased glucose

A case of falsely low blood glucose values due to a pre-analytical error.

Hyponatremia with a urine sodium measurement

A case of hyponatremia with only a urine electrolyte measurement available

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Beta-HCG’s half life

A case of rapidly decreasing b-HCG

A serum albumin of 2 g/L

A case of severely low serum albumin

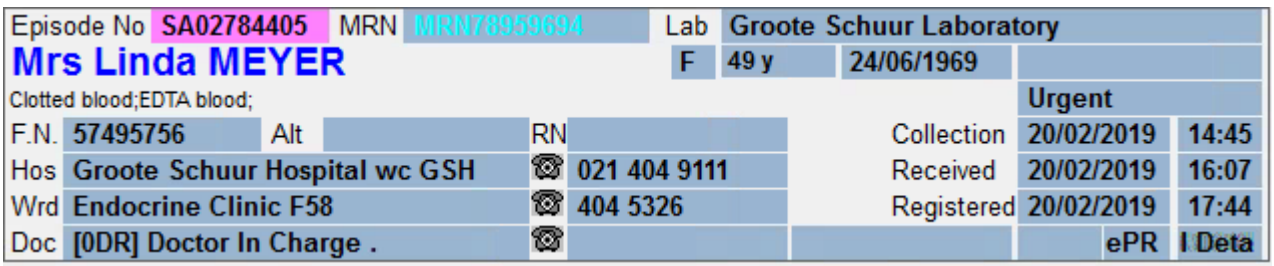
A case of elevated caeruloplasmin

A short case of elevated caeruloplasmin

Hypercalcemia with uric acid crystals

From other results it is also evident that:

HOSP #		**WARD**	Nephritic clinic
CONSULTANT	Dr. Heleen Vreede	**DOB/AGE**	49 y Female



Uric acid nephropathy with hypercalcaemia (Mrs. Linda Meyer) MRN78959694

Abnormal Result

The calcium on 20/02/2019 on bloods taken 14h45 was 3.29 (2.15-2.50 mmol/L).

Presenting Complaint

The patient presented with pain “from loin to groin” which is the typical presentation of passing a renal stone.

History

The patient has chronic renal failure (first creatinine was 362 umol/L with eGFR of 12ml/min – MDRD) on 12 December 2017. Creatinines relatively unchanged since then.

Upon re-evaluation of the case in 2020 it was seen that the baseline creatinine has risen to ~445 umol/L indicating a worsening of the chronic renal failure eGFR now 9 ml/min – by both CKD-EPI and MDRD formulas.

Examination

N/A

Laboratory Investigations

The patient is known with Hyperuricemia, first result 0.50 (0.16-0.36mmol/L) on 16 February 2018. The response to treatment appears poor due to continuing rising serum uric acid levels (considering whether the patient is on allopurinol).

2. Regarding the hypercalcemia:

Episode	SA04315821	SA03552076	SA03535628	SA02816641	SA02784405	SA02622825	SA02369770	SA02123812	SA01901592
Date	11/11/2020	11/12/2019	04/12/2019	04/03/2019	20/02/2019	12/12/2018	04/09/2018	23/05/2018	16/02/2018
Time	09:44	10:22	17:03	15:48	17:44	17:11	10:31	16:25	15:28

Na			135 L		139	138	139.000	138.000	137.000
K	5,3 H	4,7	4,8		4,8	4,5	4.320	4.400	4.780
Urea			17,2 H		14,3 H	16,2 H	11,3 H	18,8 H	17,1 H
Creat	443 H	484 H	434 H	444 H	446 H	475 H	334 H	408 H	415 H
MDRD	9	8	9	9	9	8	13	10	10
CKD-EPI	9								
Ca	**2,79 H**		**2,59 H**	**3,09 H**	**3,29 H**	**2,97 H**	**2.820 H**	**2.850 H**	**3,12 H**
Mg			0.94		1,05	1.00		1.060 H	.980
Phos			1,02		1,25	1,33	.980	1.240	1.110
PTH			**13,3 H**		4,3	4,6			

Cumulative history of UEC and CMP with PTH.
From above results a consistent hypercalcemia with a single raised PTH result can be seen – see “Final Diagnosis” and “Take Home Message” below.

Other Investigations

Uric acid crystals were seen on the urine microscopy reflecting uric acid nephropathy – a possible cause of the chronic renal failure, but I could not find any biopsy result or alternative explanation for the renal failure and assume it is uric acid nephropathy. The patient also appears to have been for a procedure at Urology (? Renal stone removal).

A serum protein electrophoresis with immunofixation (13/09/2018) showed no monoclonal peaks.

Final Diagnosis

Uric acid nephropathy with renal stones.

Hypercalcemia likely due to tertiary hyperparathyroidism.

Take Home Message

Uric acid nephropathy appears to be an uncommon cause of chronic kidney disease (ref. Up-to-date).

It should however be emphesized that clinicians consider the cause on a differential, as it is a manageable cause.

Hypercalcemia sometimes occur in Chronic Kidney Disease patients due to tertiary hyperparathyroidism. This is due to persistent hyperphosphatemia with resulting hyperparathyroidism leading to hypercalcemia (as opposed to the more commonly occuring **hypocalcemia** is renal failure).

——Commentary by Nephrologist- Dr. Erika Jones——

WRT the Uric Acid

Difficult to say if it is cause or effect of CKD. We can only really make a diagnosis of uric acid nephropathy on kidney biopsy. But it is definitely a cause that we see on occasion.

The good news is that the creatinine has remained fairly stable in the last couple of years, unlike the UA, but as kidney function deteriorates it is expected the UA will increase.

According to our buff records she had staghorn calculi and that was labelled as the cause of her CKD.

Allopurinol in CKD is challenging as it accumulates with side effects. We have had two patients with full on Steven’s Johnson Syndrome. So if she isn’t symptomatic I wouldn’t give it to her. She is recorded as having Sarcoidosis which explains the hypercalcaemia. I think this stage is too early to have tertiary hyperparathyroidism.