Short Cases in Clinical Biochemistry

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1Q

An astute orthopaedic surgeon was seeing a middle-aged man newly referred with backache. He noted pallor of the mucous membranes and requested the following biochemical investigations, which returned as follows:

Total protein	94 g/l
Albumin	34 g/l
Serum calcium	3.51 mmol/l
Serum phosphate	1.10 mmol/l

What is the diagnosis?

2Q

There was some panic on a surgical ward when the following results were phoned back from the laboratory. They were routinely taken from a patient who had had a cholecystectomy the day before, and appeared to be doing well.

Na	K	Cl	CO_2	Urea	Creatinine
117	9.6	90	14	7.7	92
mmol/l	mmol/l	mmol/l	mmol/l	mmol/l	μmol/l
				(Specimen not haemolysed)	

What is the most likely cause?

1A

Myeloma

Backache and anaemia are both common presentations of this disease, and the high serum levels of globulin and calcium make the diagnosis almost certain. A classical 'M' band was seen on electrophoresis.

2A

'Drip arm' venepuncture

The patient had an infusion of 5% glucose with potassium chloride running into a drip entering the left hand. The harrassed house surgeon had taken the blood from the left antecubital fossa. A repeat specimen from the right arm gave normal results.

3Q

A 57-year-old man was being investigated for a peripheral neuropathy. He was not diabetic, had normal renal function, and he was not obviously malnourished. Serum levels of vitamin B_{12} and folate were:

Folate	8 μg/l
Vitamin B_{12}	1475 ng/l

The B_{12} result led to a clinical and biochemical line of enquiry which revealed the diagnosis.

What did the B_{12} level mean, and what was the cause of the man's neuropathy?

4Q

A 16-year-old girl developed progressive weakness of the arms and legs, and on admission was thought to have Guillan-Barré Syndrome. Because of increasing respiratory distress she was transferred to the intensive care unit, where blood gases were performed.

pH	7.61
pCO_2	1.8 kPa
Base excess	+ 4 mmol/l
Standard bicarbonate	21 mmol/l
Actual bicarbonate	13 mmol/l
pO_2	15 kPa

What is the acid/base disturbance, and what is the likely cause?

3A

Alcoholic neuropathy

Grossly elevated serum B_{12} levels may be due to B_{12} therapy, leukaemia or hepatocellular dysfunction (the vitamin is present in very high concentrations in the liver). Subsequent liver function tests in this patient were abnormal, and it became apparent that the man was an alcoholic.

4A

Hysterical hyperventilation

The results indicate a respiratory alkalosis—entirely different from what would be expected with respiratory muscle weakness. The girl also had positive Trousseau's and Chvostek's signs (due to increased neuromuscular excitability secondary to a reduction in ionised calcium caused by the alkalosis). With sedation and firm handling the hyperventilation ceased, and she eventually made an uneventful recovery from her polyneuritis.

5Q

A 3-month-old infant with 'failure to thrive' had become non-specifically ill in the last few days. On admission the following routine area and electrolytes were returned:

Na	K	Cl	CO_2	Urea	Creatinine
125	5.9	92	17	13.5	56
mmol/l	mmol/l	mmol/l	mmol/l	mmol/l	μmol/l

What is the likely diagnosis?

6Q

A young man of 28 years was admitted for a meniscectomy, and mentioned to the houseman that he had had hepatitis as a child. The doctor ordered 'check LFTs' which came back abnormal, and remained so on repeat sampling. The operation was delayed pending a medical opinion.

Bilirubin	31 μmol/l
AST	15 U/l
Alkaline phosphatase	52 U/l

What is the likely diagnosis and how would you confirm it?

5A

Congenital adrenal hyperplasia

The results are typical of the 'salt losing' type, and the changes are identical with an Addisonian crisis. There is hyponatraemia, hyperkalaemia, metabolic acidosis and slight uraemia.

6A

Gilbert's syndrome

The physician who was consulted noted that the patient was asymptomatic and had no abnormal physical signs. He suspected Gilbert's syndrome. A differential bilirubin estimation showed that nearly all the circulating bilirubin was unconjugated, and after a 48 hour fast the total bilirubin had increased to 68 μmol/l—confirming the diagnosis. Later, the physician had a quiet word with the houseman about performing unnecessary tests! Gilbert's syndrome is due to an inherited disorder of bilirubin uptake into liver cells, and is typified by mild unconjugated hyperbilirubinaemia which is asymptomatic and increases on fasting.

7Q

A 60-year-old man was found to have myelomatosis. The following electrolyte abnormality was also noted:

Na	K	Cl	CO_2	Urea	Creatinine
125	3.3	94	22	2.5	50
mmol/l	mmol/l	mmol/l	mmol/l	mmol/l	μmol/l

What is the likely cause of this abnormality and how would you confirm it?

8Q

A middle-aged woman with diarrhoea thought that her symptoms were provoked by milk. She had a lactose tolerance test performed with the following results:

0 min	BG	4.0 mmol/l
50 g lactose given		
30 min	BG	4.1 mmol/l
60 min	BG	4.1 mmol/l
90 min	BG	4.4 mmol/l
120 min	BG	4.8 mmol/l

Is this abnormal, and if so what further steps would you take to establish a diagnosis?

7A

Pseudohyponatraemia

The high levels of paraprotein in myelomatosis may expand the plasma volume such that Na concentration falls, though the absolute Na content is the same. This effect may also be seen in severe hyperlipidaemia, and is distinguished from 'true' hyponatraemia by a normal plasma osmolality (this patient's plasma globulin level was 148 g/l and the plasma osmolality was 290 mmol/kg).

8A

Acquired lactase deficiency

A rise of less than 1 mmol/l of blood glucose is suggestive of lactose malabsorption. To exclude generalised malabsorption, a further tolerance test should be performed with 25 g glucose with 25 g galactose (the products of lactase action on lactose)—this should give a normal absorption curve. Enzyme analysis of small intestinal biopsy material, or breath hydrogen analysis after an oral lactose load, may also be used to make the diagnosis. These further investigations confirmed lactase deficiency to be present in this woman. No underlying cause could be found, but she responded well to a lactose-free diet.

9Q

A young couple were being investigated for primary subfertility. Both were clinically healthy, and the husband's semen was normal on 3 occasions. Temperature charts to demonstrate ovulation had been inconclusive. A number of blood tests were taken from the wife at one visit, and the form was marked 'one week prior to menstruation.' The results were as follows:

Progesterone	3.8 nmol/l
17β oestradiol	150 pmol/l
FSH	3 U/l
LH	2 U/l
Prolactin	1450 U/l

Why were the samples taken at this time, and what do the results mean?

10Q

This set of results came from a patient in chronic renal failure:

Na	K	Cl	CO_2	Urea	Creatinine
138	4.7	101	15	24.3	1082
mmol/l	mmol/l	mmol/l	mmol/l	mmol/l	μmol/l

Comment on the significance of the CO_2 and Cl results.

9A

Anovulation due to hyperprolactinaemia

The marked elevation in oestrogens and (particularly) progesterone which occurs in the luteal phase of the menstrual cycle can be used as a biochemical marker of ovulation. This patient's levels are in the 'pre-ovulatory' range, meaning that ovulation has not occurred at least in this cycle. The FSH and LH levels are not raised, however, excluding ovarian failure. The prolactin level is very high and this could well be causing failure of ovulation.

Hyperprolactinaemia was confirmed by cannulated samples (in case of a 'stress' result), and causes were looked for (e.g. pituitary tumour, hypothyroidism, drugs, chest trauma etc). This patient had biochemical hypothyroidism, and thyroxine treatment restored thyroid function tests, serum prolactin and fertility to normal.

10A

Uraemic acidosis

The plasma CO_2 level is low and the plasma chloride normal. The anion gap (total cations Na + K, less the total anions Cl + CO_2) is thus elevated at 27 mmol/l (it is usually about 15 mmol/l). The cause here is metabolic acidosis due to retention of unmeasured anions (e.g. sulphate, phosphate etc)—a common accompaniment of severely impaired renal function.

11Q

A 60-year-old woman had a rectal carcinoma removed surgically. There was no evidence of spread, and her LFT's at that time were normal. At follow up 3 years later however, the following results were noted:

Bilirubin	20 μmol/l
AST	23 U/l
Alkaline phosphatase	820 U/l

What is the likely cause, and what further investigations are needed?

12Q

A 47-year-old woman presented with dry eyes and pains in the knees and elbows. Amongst the investigations, the urea and electrolytes were done:

Na	K	Cl	CO_2	Urea	Creatinine
131	3.1	109	10	6.5	87
mmol/l	mmol/l	mmol/l	mmol/l	mmol/l	μmol/l

Comment on these results.

11A

Hepatic metastases

A markedly raised alkaline phosphatase, with normal or only slightly raised bilirubin and AST, constitutes 'dissociated LFT's' and is suggestive of a space occupying lesion in the liver, such as cyst, abscess or tumour. The mechanism is obstruction of a biliary radicle, causing secretion of liver alkaline phosphatase from the obstructed duct epithelium, but little or no hepatocellular damage or obstruction to outflow of bile. It is also useful in this situation to 'type' the alkaline phosphatase by electrophoresis, or assay an alternative enzyme of hepatic origin such as γ GT.

In this case a boney origin of the alkaline phosphatase was excluded by electrophoresis, and radio-isotope liver scan showed hepatic metastases, with one large deposit near the porta hepatis.

12A

Renal tubular acidosis

The patient had Sjögrens syndrome. There is an association between this and Type I (distal) renal tubular acidosis. The low CO_2 value is characteristic of metabolic acidosis, as it is too low for a respiratory alkalosis. Hyperchloraemia (with normal anion gap) and hypokalaemia are also typical. The urinary pH does not fall below 5.5 even if ammonium chloride (0.1 g/kg) is given. Nephrocalcinosis and osteomalacia may result.

13Q

A patient with Crohn's disease had the following results:

Total protein	35 g/l
Albumin	15 /l
Calcium	1.71 mmol/l
Magnesium	0.64 mmol/l
Phosphate	0.95 mmol/l
Alkaline phosphatase	60 U/l

Comment.

14Q

The following are results of a short Synacthen Test and Insulin Stress Test performed on a 49-year-old man (tests were performed at 9 a.m. on separate days).

Synacthen (250μg I/M)

0 minutes	cortisol	133 nmol/l
30 minutes	cortisol	430 nmol/l

IST (0.1 u/kg I/V)

time	blood glucose	plasma cortisol
0 mins	4.2	275
30 mins	1.3	260
60 mins	1.9	325
90 mins	2.1	330
120 mins	2.3	295

How would you interpret these?