Na 123 mmol/l, K 2.8 mmol/l, urea 1.7 mmol/l, creatinine 45 μmol/l, Li 0.5 mmol/l, glucose 3.6 mmol/l, cholesterol 2.2 mmol/l, triglycerides 0.7 mmol/l. Immunoglobulins: normal. Urine osmolality 50 mosmol/kg.

- 1. What is the diagnosis?
  - a) Dipsogenic diabetes insipidus
  - b) Nephrogenic diabetes insipidus
  - c) Cranial diabetes insipidus
  - d) SIADH
  - e) Addison's disease

### Case 9.8

A 38-year-old lady presented with pain in her fingers, particularly at night. She was a single mother with one child. She had had one miscarriage. She also suffered bad heartburn and since her teens had been prone to Raynaud's phenomenon. She took no medication, did not smoke or drink, and had no relevant family history. The appearance of her face is shown below:



A diagnosis was made. After 12 years of follow-up she complained of shortness of breath. On re-examination, she was tachypnoeic and centrally cyanosed. Her chest was clear on auscultation, but a loud second heart sound was noted. Her chest radiograph showed no signs consistent with pulmonary fibrosis. Her ECG is shown in the following figure.

- c) Increase sodium and potassium in the diet
- d) Long-acting thiazide diuretic
- e) Triamterene
- f) Regular loop diuretic
- g) Spironolactone

## Case 13.8

A 20-year-old student presented with a three-day history of increasing malaise and headache. She felt sick and had a painful neck. She was febrile. An urgent CT followed by a lumbar puncture were carried out. Both were normal, but on account of a raised ESR she was followed up in clinic. In clinic she complained of non-specific tiredness and some shortness of breath on exercise. However, her repeat ESR was normal so she was discharged from clinic. Her lassitude persisted over the next year, and she was eventually referred back with the additional symptom of pain in her legs on running. She was found to be hypertensive, with a blood pressure of 180/110 mmHg. Femoral pulses were weak, with a radiofemoral delay. An abdominal bruit was heard.

- 1. What is the most likely diagnosis?
  - a) Superinfection of congenital aortic coarctation
  - b) Giant cell arteritis
  - c) Kawasaki disease
  - d) Takayasu's arteritis
  - e) Moya moya disease
  - f) Buerger's disease
  - g) Fibromuscular dysplasia
  - h) Retroperitoneal fibrosis
- 2. Select the most appropriate investigation from the following:
  - a) Temporal artery biopsy
  - b) Renal biopsy
  - c) Aortography
  - d) Abdominal ultrasonography
  - e) Cerebral angiography
  - f) Serial blood cultures

# Case 13.9

A 20-year-old nurse presented with a two-year history of excessive thirst, polyuria and nocturia. Her weight had increased by nearly 20 kg in that period. Her development

sella meningioma. If intrinsic compression occurs, think optic nerve glioma in neurofibromatosis.

### Case 9.3

- 1. a
- 2. f

The signs initially suggest a hereditary degenerative disorder, but the myopathy does not fit. The clues are the glue-sniffer's rash from the crisp packet placement and the drunk-like appearance.

Complications of volatile substance abuse include:

- Drunk-like appearance
- Convulsions
- Cerebellar signs
- Neuropathy
- Optic atrophy
- Altered mental state
- Abdominal pain
- Haematemesis
- Aplastic anaemia
- Rhabdomyolysis
- Hypokalaemia
- Hypophosphataemia
- Acute hepatic and renal damage
- Renal tubular acidosis

### Case 9.4

- 1. c
- 2. d

The signs are bilateral and all lower motor neuron. Saddle anaesthesia with sphincter disturbance suggests sacral roots or cauda equina pathology. Conus lesions usually have mixed upper and lower motor neuron signs, e.g. an extensor plantar. Bilateral sacral plexus lesions large enough to cause this deficit are rare. Some pain would be expected with a central disc prolapse. Dysraphism refers to disorders associated with failure of fusion of the midline structure from the neural tube. Some of the associated cysts and tumours are implicated in a late progressive cauda equina syndrome on account of cord tethering. Other intramedullary tumours

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