Generalised oedema, lethargy, personality disturbance, and recurring nightmares in a young girl

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A 17 year old girl presented with a six week history of loose, yellow motions, marked leg swelling, increased shortness of breath, lethargy, and decreased exercise tolerance. She had gained 10 kg in weight in four months. Her mother commented that over the past nine months she had been somewhat depressed and withdrawn and had suffered from recurring nightmares.

Six weeks before admission, she had complained of left sided abdominal discomfort, polyuria, and haematuria and was given a one week course of co-amoxiclav. She had been otherwise fit and well. She was taking no regular medication apart from an oral contraceptive pill which had been started two years previously. She was a non-smoker and drank little alcohol.

Examination revealed generalised swelling, with marked bilateral leg oedema. Cardiovascular, respiratory, and neurological examination were unremarkable. The abdominal veins were prominent and striae were present over the lower abdomen, although she was not jaundiced and there were no other stigmata of chronic liver disease. Shifting dullness was present but no organomegaly or abdominal masses were demonstrated.

Initially, a clinical diagnosis of nephrotic syndrome was considered prompting transfer to the renal unit. Subsequently, this was excluded by a normal 24 hour urine collection for proteinuria. The results of her other screening investigations are shown in box 1.

Questions
(1) What is the likely diagnosis?
(2) What further investigations would confirm the diagnosis?
(3) What treatment options might you consider and how would you manage this young girl?

Box 1: Screening investigations
- Full blood count: haemoglobin 106 g/l, leucocyte count 12.0 x 10⁹/l, platelets 92 x 10⁹/l.
- Urea and electrolytes: sodium 130 mmol/l, potassium 2.4 mmol/l, urea 3.1 mmol/l, creatinine 76 µmol/l.
- Erythrocyte sedimentation rate and C reactive protein: normal.
- Liver function tests: albumin 20 g/l, bilirubin 51 µmol/l, alkaline phosphatase 208 IU/l, aspartate aminotransferase 131 IU/l.
- Prothrombin time: 45.2 sec.
- Kaolin cephalin clotting time: 99.7 sec (control 39).
- Serum immunoglobulins: polyclonal IgG rise.
- Autoantibody profile: weakly positive smooth muscle antibody (1:10); antinuclear and antimitochondrial antibodies negative.
- Serum α-fetoprotein and α₁-antitrypsin levels normal.
- Serum B12 1668 ng/l (120–600).
- Serum folate 7.2 µg/l (1.6–6.0).
- Serum ferritin 281 µg/l.
- Serum copper 0.3 mg/l (0.7–1.6).
- Serum caeruloplasmin 0.06 g/l (0.2–0.45).
- Stool, urine, and ascitic fluid culture: normal.
- Viral titres and hepatitis serology: negative.
- Ascitic fluid: protein 1.2 g/l, glucose 5.4 mmol/l, leucocytes <100/mm³.
- Abdominal ultrasound: normal liver, spleen, pancreas, and kidneys. Gross ascites and a few dilated, fluid filled bowel loops. Doppler studies indicate normal flow through portal veins. No evidence of Budd-Chiari syndrome.
An unusual pituitary mass presenting with panhypopituitarism and hyponatraemia

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A 72 year old Asian man was transferred to our institution for work-up of hyponatraemia and an intrasellar mass. At an outside hospital, the patient presented with a fever of 40°C (104°F) and mental status changes. The patient had been exposed to ill children and reported symptoms of upper respiratory tract infection a week before presentation. On admission to the outside hospital, he had a serum sodium of 133 mmol/l which decreased to 125 mmol/l with onset of mental confusion. Lumbar puncture findings were normal. Computed tomography of the head done at that point showed a 2.1 cm sellar mass with suprasellar extension, elevating the optic chiasm (fig 1). His fever resolved after treatment with azithromycin. His mental status markedly improved after intravenous infusion of saline and a single dose of hydrocortisone 100 mg given intravenously.

On admission to our hospital, his pulse was 80 beats/min, blood pressure 140/88 mm Hg, temperature 37.3°C, and respiratory rate 20 breaths/min. He was alert and oriented. Systemic examination, including heart, lungs, abdomen, and central nervous system was normal. The neuro-ophthalmic evaluation revealed bitemporal visual field defects with no papilloedema. The endocrine evaluation showed marked abnormalities. Free testosterone was less than 4.9 nmol/l (normal range 76.3–721.1 nmol/l), free and weakly bound testosterone was less than 0.12 nmol/l (2.1–20.1 nmol/l), total testosterone was less than 0.35 nmol/l (7.3–26.0 nmol/l), sex hormone binding globulin was 29 nmol/l (13–71 nmol/l). Luteinising hormone was 0.9 IU/ml (2.0–6.0 IU/ml), follicle stimulating hormone 1.6 IU/ml (1.0–15.0 IU/ml), random cortisol <28 nmol/l (55.2–607.0 nmol/l), total triiodothyronine 0.8 nmol/l (1.2–2.5 nmol/l), total thyroxine 63.1 nmol/l (64.4–154.4 nmol/l), and thyroid stimulating hormone 0.26 mU/l (0.2–5.0 mU/l). His serum prolactin concentration was 9 µg/l (2.0–18.0 µg/l); serum growth hormone was not measured. Serum sodium on admission was 132 mmol/l (136–146 mmol/l), potassium 4.2 mmol/l (3.5–5.3 mmol/l), blood urea nitrogen 6.4 mmol of urea (7.0–22 mmol of urea), and creatinine 97.2 µmol/l (61.9–132.6 µmol/l). Prostate specific antigen was undetectable at 0.

The patient underwent transsphenoidal hypophysectomy with complete removal of the tumour. During the surgery the tumour could be removed easily and was not attached to the dura. The surgical specimen showed a 2.3 × 1.7 × 0.4 cm tumour. The pathology revealed fascicular arrangement of spindle shaped cells with cigar and cigarette shaped nuclei (figs 2 and 3). The tumour cells were S-100 stain positive. Other special stains for prolactin, growth hormone, adrenocorticotrophic hormone, epithelial membrane antigen (EMA), as well as glial fibrillary acidic protein were negative. Electron microscopy showed evidence of basal laminae.

Postoperatively, he is doing well on replacement doses of levothyroxine, cortisone, and testosterone. Serum sodium was within the normal range on follow up laboratory analyses. Postoperative magnetic resonance imaging did not reveal any tumour, but did show postoperative changes in the pituitary fossa. Follow up in the ophthalmology clinic did not reveal any visual field defects.
Questions
(1) What is the differential diagnosis of an intrasellar mass?
(2) What is the cause of the intrasellar mass in this patient and what are its diagnostic features?
(3) What is the cause of hyponatraemia in this patient?

A Mauritian woman with fever, abdominal pain, and facial palsy

P Gyawali, D Agranoff, D C Macallan

A previously well 43 year old woman of Mauritian origin presented to the surgeons with abdominal pain, nausea, and vomiting occurring over a period of eight weeks. She described it as a band-like constricting sensation encircling the upper abdomen and lower chest. Six weeks previously she had suffered a right lower motor neurone facial nerve palsy, which had resolved spontaneously. There were no respiratory symptoms. She had been exposed to tuberculosis at the age of 2 before immigrating to the UK. She was on no regular medication, and did not smoke or drink alcohol.

On examination she had a low grade fever and was tender in the epigastrium. There was no obvious BCG scar. She had altered sensation to light touch and pinprick in the lower thoracic and upper abdominal dermatomes. She had modestly deranged liver function tests: bilirubin 9 µmol/l, alanine transaminase 86 U/l, alkaline phosphatase 321 U/l, and amylase 417 IU/l but her full blood count and urea and electrolytes were normal.
She was admitted for further investigations. Five days after admission she complained of weakness in the left leg and was found to have impairment to light touch and pinprick sensation over the knee with loss of the left knee reflex. This was rapidly followed by development of a left lower motor neurone VIIth nerve palsy on the opposite side to the previous lesion. In view of her past exposure to tuberculosis and persisting low grade fever, she was transferred to the care of the infectious diseases team for further evaluation.

Chest radiography showed paratracheal lymphadenopathy but induced sputum was negative for acid-fast bacilli and she had no reaction to a tuberculin skin test (10 units). Percutaneous liver biopsy was undertaken and histology is shown in fig 1. A magnetic resonance scan of the spine is shown in fig 2. An ultrasound scan showed diffuse echogenic abnormality in the liver.

Questions
(1) What is the likely diagnosis and what supportive investigations would be helpful?
(2) What further test should be performed to help guide management?

Hyperplastic polyposis coli associated with dysplasia

M A Whittaker, N J Carr, N Cripps

Case reports

CASE 1
A 56 year old woman with no previous history of gastrointestinal disease presented with a short history of abdominal pain, altered bowel habit, weight loss, and anaemia. There was no mucus or blood in the stool. Colonoscopy revealed a constricting carcinoma at the hepatic flexure of the transverse colon and numerous sessile and pedunculated polyps both proximal and distal to the tumour. Subtotal colectomy with ileostomy anastomosis was performed. The resection specimen showed a Dukes’ B adenocarcinoma with vascular invasion and contained 53 polyps, two of which measured 30 mm in diameter and many measured around 15 mm; two further polyps were sampled from the residual colon. Histologically these all proved to be hyperplastic polyps with two polyps showing foci of low grade dysplasia. The patient is still being followed up and 10 further small polyps (largest 8 mm) were found at colonoscopy six months postoperatively. She is a member of a large family and so far two first degree relatives have undergone colonoscopy. A 25 year old daughter had two hyperplastic polyps of the right colon; a second 28 year old daughter had no polyps.

CASE 2
A 33 year old man presented with a perianal abscess. Examination under anaesthesia performed at the time of drainage demonstrated irregularity and thickening of the rectum, clinically suspected to be a carcinoma. Biopsy specimens, however, showed a large hyperplastic polyp with epithelioid granulomata within the lamina propria suggestive of Crohn’s disease. In total, 29 polyps were found at subsequent colonoscopy with the largest measuring 25 mm in diameter. Twenty four polyps were sampled and all showed hyperplastic morphology but three polyps also showed foci of mild dysplasia. There was no evidence of invasive tumour. He was treated with mesalazine for his suspected Crohn’s disease and is currently well, although with several residual left sided polyps. He is being regularly followed up with colonoscopy.

Questions
(1) What is the differential diagnosis?
(2) What is the diagnosis for these patients?
(3) How would you manage patients with this diagnosis?
Confusion in an elderly patient: an uncommon diagnosis for such a common event

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A 90 year old woman was admitted to the emergency department with delirium and falls. She had a recent history of cognitive impairment, insomnia, drowsiness, and progressive disability. Three months before admission, in the course of a medical check-up, an isolated increased plasma thyroid stimulating hormone (TSH 8 mIU/l, normal values 0.5–2.9) was discovered, leading to a levothyroxine therapy (50 µg daily). This patient was previously alert and able to live almost independently. Physical examination was unremarkable, except for fidgeting and restlessness. Other biochemical investigations were normal. A cranial tomographic study was obtained and revealed no intracranial haematoma but unexpectedly showed a pituitary macroadenoma, subsequently confirmed by magnetic resonance imaging (fig 1).

Questions
(1) What is the initial diagnosis?
(2) In view of the results, which diagnosis should be considered and which parameters shall be measured?

Figure 1  Magnetic resonance imaging of the brain shows a large pituitary adenoma (arrows).
Siblings with multiple soft tissue calcifications

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Two brothers aged 10 years and 18 years presented with multiple soft tissue calcareous swellings around the elbows, arms, knees, and forearms, which had been present for the last three years. Both were in good general health and there was no history of trauma. On examination, the younger boy had a calcified soft tissue swelling around the left knee joint with a sinus around the medial side of the knee. There was occasional discharge of white chalky material from the sinus. Movements at the knee were full and there was no neurovascular problem. The other brother had calcareous, firm, soft tissue swellings over the right lower thigh, left and right arms, and around both the elbow joints. Movements at the elbow and knee joints were normal and he had no discharging sinus. Their serum calcium, serum phosphorus, alkaline phosphatase, and urinary calcium were within normal limits. The erythrocyte sedimentation rate and the leukocyte count were normal. A test for lupus erythematosus was negative and there was no serum rheumatoid factor. Serum uric acid was normal. Plain radiographs of the elbows, knees, and thigh were taken; those of the knee in the younger brother and elbow and arm in the older brother are shown in figs 1–3.

Questions
(1) What are the radiological findings?
(2) What is the diagnosis?
(3) What is the pathogenesis and treatment of this condition?
A man with a murmur requiring nutritional support

S D Hearing, I S Shaw, A Day, C S J Probert

A 52 year old man complained of severe abdominal pain of 24 hours’ duration. He had had two episodes of bloody diarrhoea in the past six hours. Medical history was unremarkable. On examination he was systemically unwell, pyrexial (37.5°C), and was noted to have several splinter haemorrhages in his fingernails. Cardiovascular examination revealed a tachycardia (120 beats/min), an irregularly irregular pulse, blood pressure 100/80 mm Hg, and a diastolic murmur loudest at the apex. Abdominal examination revealed generalised tenderness with guarding in the lower abdomen. Initial investigations were as follows: haemoglobin concentration 113 g/l, white cell count 21 × 10⁹/l, platelet count 550 × 10⁹/l, serum sodium 134 mmol/l, potassium 3.4 mmol/l, urea 9.2 mmol/l, creatinine 102 µmol/l, arterial pH 7.22, and bicarbonate 14 mmol/l.

At laparotomy a large amount of small and large bowel was resected, leaving 20 cm of jejunum, and a jejuno-transverse colonic anastomosis was performed.

Three months after surgery the patient was admitted with severe dehydration, anuria, and carpopedal spasm. His serum sodium concentration was 127 mmol/l, potassium 3.3 mmol/l, urea 58 mmol/l, and creatinine 403 µmol/l.

Questions

(1) At presentation what diagnosis would you consider in this patient and how does this relate to the findings on examination of the cardiovascular system?

(2) How are feeding and fluid requirements assessed in patients who have undergone intestinal resections and what plans should be made for nutritional support for this patient after laparotomy?

(3) What has happened to the patient three months after surgery and what caused the carpopedal spasm?

Heart failure, a thick tongue, and an abnormal cranial computed tomogram

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Answers on p 545.

A 21 year old man was referred by primary care physicians with complaints of attacks of thick tongue lasting few minutes, of one year’s duration. The attacks had increased in frequency over the previous month. He also complained of lethargy, abdominal pain, intermittent dysphagia, breathlessness on accustomed exertion, and mild headache of the same duration. On examination he was of average build and nourishment, and was breathless at rest. His pulse rate was 110 breaths/min, with a low pulse volume. Blood pressure was 80/70 mm Hg. He had raised jugular venous pressure, pedal oedema, tender hepatomegaly, and ascites. There was cardiomegaly and murmur of tricuspid regurgitation. On active movements of the tongue it became broad, thick, and stiff; and came back to normal after few minutes. The rest of the general and systemic examinations was normal. An earlier non-contrast enhanced cranial computed tomogram is shown in fig 1.

Questions

(1) What is the underlying condition, which can explain his thick tongue and cranial computed tomogram abnormalities?

(2) What is the pathophysiology of his cardiac failure?

(3) What are the other cardiovascular manifestations of this condition?
Massive pleural effusion

A Salih

A 50 year old alcoholic male patient, with a known history of chronic pancreatitis and insulin dependent diabetes but no past history of respiratory problems, presented with progressive shortness of breath for a duration of three months and epigastric discomfort for two days before admission.

On examination he was pyrexial, dyspnoeic, and tachypnoeic with dullness and reduced air entry of the left chest. The abdomen was soft, with slight epigastric tenderness on deep palpation.

His white cell count was $23.5 \times 10^9/l$. Serum amylase was 2500 IU/l, serum bilirubin 81 $\mu$mol/l, and alkaline phosphatase 1242 IU/l. Pleural aspiration yielded a bloodstained exudative fluid with a protein concentration of 56 g/l.

Chest radiography revealed left sided massive pleural effusion (fig 1). Computed tomography demonstrated pancreatic ductal and parenchymal calcification (fig 2). Previous computed tomography performed a year before revealed a pseudocyst arising from the head of pancreas, adjacent to the left lobe of the liver (fig 3). The cyst had settled with conservative management.

Questions

(1) What is the most probable cause for the massive pleural effusion?
(2) How should the diagnosis be confirmed?
(3) What would be the differential diagnosis?
(4) What are the options for treatment?

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