An unusual presentation of a common disorder

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A 59 year old right handed man presented with a one week history of twitching of the right hand side of his face. This would spread to involve his right arm and hand. There was no background medical history of note and he was on no medication. On examination the right hand side of his face was continually twitching. Neurological examination did not reveal any further abnormalities. The results of investigations are shown in box 1.

Questions
(1) What is this uncommon epilepsy syndrome called?
(2) What is the connection between the abnormal biochemistry and the neurological presentation?
(3) In what proportion of cases of this syndrome will computed tomography or magnetic resonance imaging of the brain be abnormal?

Young male with recurrent flaccid quadriplegia and complete recovery

A S Kashyap, S Kashyap

A 25 year old male shopkeeper was brought to the emergency room with sudden onset flaccid quadriplegia of five hours’ duration. The proximal muscle groups were weaker than the distal. He gave a history of similar episodes for the last four years. The episodes usually followed a period of rest after moderate to severe exertion. Each attack lasted three to 10 hours, and was followed by spontaneous complete recovery. The frequency varied from once per day to three times per week. There was no history of loss of consciousness, diaphoria, drop attacks, dysarthria, dysphagia, alcohol, or drug intake. There was no history suggestive of hyperthyroidism. His parents and two sisters denied any history of similar neurological illness or hyperthyroidism. Clinically he had flaccid motor paralysis with absent deep tendon jerks. There was mild wasting of the thigh and arm muscles. Cranial nerves, fundi, higher mental functions, and sensations were normal. There was no goitre, tachycardia, tremor, or weight loss. Abnormal investigations at admission are listed in box 1.

Blood counts, creatinine, serum sodium, chloride, bicarbonate, calcium, glucose, cortisol, and supine aldosterone concentrations were normal. Nerve conduction velocities were normal.

Questions
(1) What is the diagnosis?
(2) What is the basic pathophysiology of this disorder?
(3) How will you prevent further attacks?
Swelling in the caesarean section scar

M Hussien, J W R Peyton

A 38 year old women, para 3+1, presented with a two month history of painful swelling in her caesarean section scar. She had two caesarean sections seven and 10 years earlier. She had a long history of irregular menstrual periods.

On examination there was a tender, firm 2 × 2 cm swelling in the right side of the phannensteil incision scar. There was no cough impulse.

Questions
(1) What is the differential diagnosis?
(2) What investigations should you consider?
(3) How would you treat this patient?

An unusual case of chorea gravidarum

A Qasim

A 24 year old right handed Pakistani woman who was eight weeks' pregnant presented with a two day history of involuntary movements of her left arm, hand, and foot. There was no significant past medical history or family history, and she was taking no regular medication. On examination there were choreiform movements of the left upper limb and left foot. Her speech was normal and there was no other neurological abnormality. She was apyrexial, and examination of the cardiovascular, respiratory, and abdominal systems was unremarkable. Fundoscopy and slit lamp examination of the eyes was normal. Full blood count, urea and electrolytes, liver function tests, and thyroid function tests were normal. She had a polyclonal rise in IgG, with no evidence of autoantibodies. C reactive protein was <6 mg/l and the antistreptolysin O titre 320. Electrocardiography and echocardiography showed no abnormalities. Obstetric ultrasound was performed which showed a viable fetus with estimated gestational age of 10 weeks. Magnetic resonance imaging (MRI) of the brain was normal. She was treated with haloperidol 1 mg three times daily and her chorea improved markedly over the next three days.

On day 8 she suffered a primary focal seizure with violent movements of the right arm leading to a secondary generalised seizure. Emergency computed tomography of the brain with and without contrast showed no abnormality. Cerebrospinal fluid obtained by lumbar puncture was sterile with normal cell counts. An electroencephalogram was asymmetrical with generalised excess of left sided theta and delta waves unaffected by photic stimulation. In the 24 hours after the seizure she developed expressive dysphasia, right sided hemiplegia, and worsening left sided chorea. MRI of the brain was performed on day 10 (fig 1). She was anticoagulated with intravenous unfractionated heparin, and within six hours there was an increase in right sided power to grade 4 out of 5 and improvement of her dysphasia and chorea. After bleeding from her vagina on day 27 miscarriage was diagnosed on obstetric ultrasound. She was discharged after 35 inpatient days: she had mildly reduced right sided power (grade 4 out of 5) and mild dysphasia. Life long treatment with warfarin is planned and she has been advised against future pregnancies.

Questions
(1) Give a differential diagnosis of chorea in pregnancy
(2) Describe the abnormalities in the MRI image (fig 1)
(3) What underlying causes should be sought in this patient?
A 72 year old man with manic depression, who had recently started lithium treatment, presented to casualty with a two day history of severe nausea and vomiting. His serum urea and electrolytes were normal (sodium 139 mmol/l, potassium 4.1 mmol/l, creatinine 94 mmol/l, and urea 5 mmol/l) and he was discharged from the accident and emergency department with a presumptive diagnosis of gastroenteritis.

He represented 12 hours later with persistent vomiting and oliguric prerenal failure initially thought to be secondary to lithium toxicity. There was only minimal tenderness in the epigastrium but no signs of peritonism or abdominal masses; his pulse was 90 beats/min and blood pressure 105/50 mm Hg without a visible jugular venous pressure. His blood urea was now 28 mmol/l, sodium 141 mmol/l, potassium 4.3 mmol/l, and creatinine 310 mmol/l, with normal full blood count, blood gases, creatinine kinase, amylase, urine microscopy, a subtherapeutic lithium concentration 0.35 mmol/l (normal range 0.4–0.8 mmol/l), and an electrocardiograph showed sinus tachycardia only. His central venous pressure was low (−2 mm Hg) and he was resuscitated with fluids. Chest radiography was performed (figs 1 and 2) and a nasogastric tube was inserted but with some difficulty, draining only 100 ml of gastric juice. Twelve hours later his symptoms still had not improved. His renal function was unchanged and his vomitus had now become blood stained.

Questions
(1) What do the chest radiographs show?
(2) What single diagnostic investigation is required?
SELF ASSESSMENT ANSWERS

An unusual presentation of a common disorder

Q1: What is this uncommon epilepsy syndrome called?
Epilepsia partialis continua (EPC).

Q2: What is the connection between the abnormal biochemistry and the neurological presentation?
Hyperglycaemia is a well recognised metabolic cause of EPC.¹

Q3: In what proportion of cases of this syndrome will computed tomography or magnetic resonance imaging of the brain be abnormal?
50%-60%.

Discussion

EPC is defined as clonic muscular twitching repeated at fairly short intervals in one part of the body for a period of days or weeks. It is a form of focal status epilepticus manifesting as continuous focal motor seizures.² It is associated with a wide range of electroencephalographic abnormalities including focal spikes and focal slow waves². However in some cases the electroencephalogram may be normal.¹ Causes of EPC include structural abnormalities such as central nervous system tumours, trauma, cerebral infarction, intracerebral haemorrhage, cerebral abscess, neuronal migration disorders, and vascular malformation.² However in up to 50% of cases conventional brain imaging may be normal.³ Causes where brain imaging may be normal include hypocalcaemia, hyperglycaemia, hepatic encephalopathy, and encephalitis.²

The association between hyperglycaemia and EPC is well recognised. In some cases patients have an underlying structural abnormality such as cerebral infarction. In others hyperglycaemia can cause EPC in the absence of any neurological disease. In these patients treatment of the seizures and reversal of the metabolic disorder prevents any long term neurological sequelae.¹ In the majority of patients with EPC associated with hyperglycaemia EPC occurs before impairment of consciousness and coma; it is important therefore to recognise this association. All patients presenting with EPC should have immediate determination of blood glucose concentrations. This could lead to the detection of previously undiagnosed diabetes mellitus and prompt reversal of the neurological condition. Our patient was treated with insulin, intravenous fluids, and a carbamazepine 400 mg daily. His seizures quickly resolved and he has not had any recurrence. Neurological examination at a follow up neurology outpatient clinic three months later did not reveal any abnormalities.

Final diagnosis
Epilepsia partialis continua, secondary to hyperglycaemia.

Young male with recurrent flaccid quadriparesis and complete recovery

Q1: What is the diagnosis?
The diagnosis is thyrotoxic hypokalaemic periodic paralysis (THPP). This rare condition is seen in young Latin American and East Asian men with thyrotoxicosis. The clinical features of thyrotoxicosis are usually not apparent. The human leucocyte antigen types A2 Bw22 and Aw19 B17 increase the relative risk. The age of onset is usually before 30 years, and familial occurrence is unusual. The frequency of attacks varies from daily to yearly, and each attack lasts two to 12 hours. The attacks are precipitated by a high carbohydrate diet, sodium intake, emotional stress, and strenuous exertion followed by rest/sleep. It never occurs during vigorous physical activity. The higher mental functions and sensations are not affected. It is characterised by episodic, sudden onset, flaccid quadriparesis, with absent or hypoactive reflexes. The proximal limb weakness is more than the distal, and lower limbs are more prominently involved. The ocular, bulbar, and respiratory muscles are rarely involved. It responds to potassium administration. The frequency and severity of THPP episodes is reduced by β-adrenergic blockers, antithyroid drugs, and by avoidance of strenuous physical activity and a high carbohydrate diet. Rendering the patient euthyroid abolishes this condition. In the absence of effective treatment chronic progressive interattack weakness may develop. The muscle biopsy specimen often shows vacuolar myopathy.¹

Q2: What is the basic pathophysiology of this disorder?
The basic pathophysiology in THPP is the development of hypokalaemia due to an intracellular shift of potassium. The total body potassium is normal. The cause of this shift is uncertain. Molecular defects in skeletal muscle calcium channels, leading to a decrease in activity of the calcium pump have been identified.¹ Calcium transport disturbances may adversely change muscle excitation-contraction coupling and account for acute paresis in THPP. The contractile apparatus is normal. Effects of insulin on potassium uptake in muscle suggest that an abnormality of muscle membrane may be involved. Increased
activity and numbers of Na+, K+-ATPase channels in skeletal muscle and platelets have been identified in hyperthyroid patients with or without periodic paralysis. Hyperthyroidism leads to increased β-adrenergic receptor responsiveness and this also contributes to increased activity of Na+, K+-ATPase channels. This may explain the mechanism of β-blockers in prevention and of the hyperadrenergic state of exercise in provocation of THPP attacks. Hyperinsulinaemia is present in many patients with THPP and increases the Na+, K+-ATPase induced transport of potassium. This is important in the precipitation of paralytic attacks and may underlie the provocation of paralysis with high carbohydrate intake.

Q3: How will you prevent further attacks?
Establishment of the euthyroid state will abolish the attacks. β-Adrenergic blocking agents are useful for reducing the frequency and severity of attacks while measures to control thyrotoxicosis are instituted. Strenuous exertion and high carbohydrate diet avoidance also helps. Acetazolamide is not useful in attack prevention.

Follow up
This patient was being managed as a hysterical conversion reaction by primary care physicians. Once the diagnosis of THPP was established the patient was managed with intravenous potassium supplements under electrocardiographic monitoring. Six hours later his paralysis resolved and serum potassium concentrations rose to 4.1 mmol/l. Subsequently he was put on propranolol 80 mg/day and carbimazole 30 mg/day. Six weeks later his thyroid function tests were normal. There has been no recurrence of paralytic attacks over a follow up of nine months.

The patient refused radioiodine therapy, and is on carbimazole and propranolol.

Learning point
It is essential that THPP be excluded in acute pure motor flaccid hyporeflexic quadriparesis with clear sensorium, particularly in young men.

Swelling in the caesarean section scar
Q1: What is the differential diagnosis?
The differential diagnosis of this swelling with late onset after surgery include organised haematoma, stitch abscess granulomatosis, incisional hernia, endometrioma, and neoplasm which is either intra-abdominal or neoplasm of the abdominal wall itself as sarcoma, desmoid tumour, or metastatic cancer.

Q2: What investigations should you consider?
An ultrasound scan will help to determine if the mass is cystic or solid and exclude underlying intra-abdominal pathology. This was done (fig 1) and showed a 2 cm hyperechoic area in the abdominal wall musculature with no posterior defect, which excluded the possibility of incisional hernia. There was no associated intra-abdominal pathology.

Other investigations which could be considered include computed tomography and fine needle aspiration cytology.

Q3: How would you treat this patient?
Surgical exploration of the scar and excision of the swelling should be considered. This was performed and there was a 2 × 2 cm firm mass in the subcutaneous tissue which was extremely adherent to the anterior rectus sheath. The sheath was partially excised to ensure complete removal of the mass. Histopathology examination confirmed the diagnosis of a desmoid tumour.

Final diagnosis
Thyrotoxic hypokalaemic periodic paralysis.


Figure 1 Ultrasound scan showing 2 × 2 cm hyperechoic area in the abdominal wall musculature with no posterior abdominal wall defect.
tion of the resected tissue confirmed the presence of endometriosis.

Discussion
The cause of surgical scar endometriosis is believed to be iatrogenic transplantation of endometrium into the surgical wound particularly during late pregnancy and delivery where endometrial tissue has the maximum potential for ectopic implantation. The reported incidence after term or near term caesarean sections ranges from 0.1%–0.4%.

The endometrial tissue inoculum is subsequently stimulated by oestrogens to proliferate until it becomes large enough to cause symptoms. The time interval between surgery and the onset of symptoms is variable and ranging from a few months to several years with an average interval of 4.5 years. Incisional endometriosis usually occurs in the right side of the scar as it is the usual side of the operator and it is densely attached to fascia.

The typical presentation is a tender mass adjacent to surgical scar of gynaecological procedure. The tenderness is usually intermittent and is associated with the menstrual cycle, but this pattern is not always present. The rare incidence and late onset with slow and intermittent progression of symptoms after surgery is usually misleading and is the main cause of misdiagnosis. It is also interesting to note that incisional endometriosis is rarely found in association with symptoms or findings of pelvic endometriosis.

Medical treatment of surgical scar endometriosis is similar to treatment of other forms of endometriosis but it only produces temporary alleviation of symptoms followed by recurrence after cessation of the treatment. Therefore, surgical excision is the treatment of choice and should include the mass and a clean surgical margin which may include part of the rectus sheath in order to prevent recurrence. Synthetic mesh may be necessary to close large defects of rectus sheath. A preoperative period of suppression of ovulation may be considered to reduce vascularity of the lesion, which may facilitate surgery and reduce postoperative complications.

Final diagnosis
Incisional endometriosis.

An unusual case of chorea gravidarum
Q1: Give a differential diagnosis of chorea in pregnancy
Although approximately half the cases of chorea gravidarum are idiopathic, rheumatic fever and antiphospholipid syndrome underlie most of the remainder. It is also important to consider Wilson’s disease, thyrotoxicosis, Huntington’s disease, subcortical infarction, and drug induced choreas.

Q2: Describe the abnormalities in the MRI image
There is extensive cerebral infarction in the middle cerebral artery territory.

Q3: What underlying causes should be sought in this patient?
In a young patient with cerebral infarction, in the absence of hypertension and atrial fibrillation, vasculitides and thrombophilic tendencies must be considered. This patient was normotensive and in sinus rhythm, with no increase in C reactive protein or inflammatory markers. Testing for thrombophilia showed no anticardiolipin antibody, normal antithrombin III levels, normal prothrombin gene, mildly reduced protein S, and activated protein C resistance. She was homozygous for factor V Leiden.

Discussion
Chorea gravidarum is a rare condition which usually presents in the first trimester of pregnancy with bilateral involuntary movements, often with slurred speech and altered affect. It is almost always self-limiting and resolves when the pregnancy ends either with delivery, miscarriage, or termination. About half the cases are idiopathic, with rheumatic fever and antiphospholipid syndrome (APLS) underlying most of the remainder. It is also important to consider Wilson’s disease, thyrotoxicosis, Huntington’s disease, subcortical infarction, and drug induced choreas. MRI in both primary and secondary cases is usually entirely normal, though there may be subtle subcortical changes and infarction. Treatment of the underlying cause usually leads to resolution of the chorea. Recurrences may occur in subsequent pregnancies, particularly if APLS is the cause. Dopamine antagonists haloperidol and chlorpromazine are useful in symptom control and are seen to be safe when used in low dose.

This young woman developed rapidly progressing neurology and has a thrombophilic tendency as the underlying cause of her extensive cerebral infarction. Anticoagulation with intravenous heparin arrested the deterioration and her neurological abnormalities improved, leaving her with mild impairment. There is an association between activated protein C resistance and spontaneous abortion, which is likely to be due to placental infarction. Although 5%–10% of Europeans are heterozygous for factor V Leiden, the prevalence in Asians is less than 0.5% and reports of arterial thrombosis in Asians due to homozygosity are rare. This is the first case of chorea gravidarum and...
progressive cerebral infarction due to factor V Leiden homozygosity in the literature, and there was a good clinical response to treatment with unfractionated intravenous heparin.

Final diagnosis
Chorea gravidarum and extensive cerebral infarction secondary to underlying homozygosity for factor V Leiden.


Learning points
- Nausea and vomiting should always be taken seriously.
- Borchardt’s triad of constant epigastric pain, nausea/vomiting and difficulty in passing a nasogastric tube should suggest the presence of acute gastric volvulus.
- Chest radiograph usually shows the presence of paraoesophageal hernia with two air fluid levels: one beneath the left hemidiaphragm and the other in the retrocardiac mediastinum.
- Gastroscopy is useful in the initial diagnosis and the treatment but surgery still remains the definitive treatment.

Nausea and vomiting, a cause for concern?

Q1: What do the chest radiographs show?
The chest radiographs appear to show a hiatus hernia with a fluid level (fig 1, arrowed) but no free gas under the diaphragm or intramural air.

Q2: What single diagnostic investigation is required?
An urgent gastroscopy should be performed.

Passage of the endoscope through the oesophagus was achieved without difficulty. Upon entering the proximal stomach, the mucosa appeared normal; the lumen was not readily visualised and a large acute gastric volvulus with a paraoesophageal hernia was diagnosed. The endoscope was carefully manipulated through the narrowed lumen formed by the twisted gastric folds of the fundus and advanced into the distal body and antrum. The volvulus was decompressed aspirating 1.2 litres of bloodstained fluid and then subsequently reduced with the gastroscope. Thereafter, within 15 hours the patient’s symptoms cleared and his renal function was back to normal by which time he no longer required further resuscitation from intravenous fluids.

He later went on to have an elective fundoplication of the paraoesophageal hernia; this prevents further recurrence of the gastric volvulus.

Discussion
Acute gastric volvulus is unusual, with over 300 cases being reported since the initial description by Berti in 1866. It is seen in all ages with a peak incidence in the fifth decade. The mortality rates are high mainly from gastric infarction, varying from 30% to 50%. Unfortunately this often reflects the delay in diagnosis and treatment particularly when gastric volvulus is not considered.

The classic Borchardt’s triad of nausea, vomiting, severe constant chest or epigastric pain, and difficulty in passing a nasogastric tube should raise the suspicion of acute gastric volvulus particularly when there is an associated paraoesophageal hernia. The acute form is seen less often than the chronic (almost half of cases). Seemingly vague symptoms of nausea and vomiting may be one of the presenting complaints and can surprisingly be overlooked, so making the diagnosis difficult. Our case shows this point well, where the volvulus was camouflaged by renal impairment presumed initially to be secondary to lithium toxicity. An erect chest radiograph should give clues to the diagnosis also, for example, the presence of double air fluid levels—beneath the left hemidiaphragm and in the retrocardiac mediastinum as described by Rosselet.4

Gastric volvulus presents acutely with closed loop obstruction (which contributed towards this patient’s renal impairment), strangulation, ischaemia, or perforation of gastric viscus. These patients have rotations greater than 180 degrees in the cardiophrenic or mesenteroaxial plane. Vascular compromise leads to gastric gangrene in 5% to 28% of patients presenting with acute gastric volvulus.5

The definitive treatment of patients with gastric volvulus has been surgical to reduce the volvulus, thereby correcting the underlying pathological feature.6 If gastric necrosis is found, the stomach is repaired by local excision, subtotal or even total gastrectomy. Anterior gastropexy in which the greater curve of the stomach is fixed to the undersurface of the anterior abdominal wall is performed to prevent recurrence.

Non-surgical correction of acute gastric volvulus is also of benefit as it allows time for elective surgical repair so as to avoid gastric ischaemia. This includes nasogastric decompression or endoscopic reduction. The latter technique involves the formation of a loop in the form of an alpha, further advancement of the endoscope tip into the antrum of the stomach, and uncoiling of the “alpha loop” with subsequent reduction of the gastric volvulus.7

Acute gastric volvulus must therefore be considered early in the differential diagnosis of nausea and vomiting, especially after a normal clinical examination.
We believe this is under-recognised at initial presentation because the symptoms are not taken seriously and perhaps a little underestimated.

**Final diagnosis**

Acute gastric volvulus with paraoesophageal hernia.


Young male with recurrent flaccid quadriparesis and complete recovery

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