Myotonia dystrophica – first presentation as severe left ventricular failure complicating dilated cardiomyopathy

Sir,

Myotonia dystrophica is an autosomal dominant multi-system disorder which in adults affects the specialized cardiac conduction tissue frequently, but the myocardium itself only rarely clinically,\(^1\) although electron microscopic studies show early myocardial involvement.\(^2\) Cardiomyopathy has been described,\(^3,4\) but we are unaware of reports of first presentation of myotonic dystrophy with dilated cardiomyopathy and left ventricular failure.

A previously well, 30 year old infertile man presented with breathlessness at rest and paroxysmal nocturnal dyspnoea. There was no significant family or drug history. He was orthopnoeic and hypotensive (blood pressure 90/70 mmHg) with a raised jugular venous pressure. His pulse was 112/min and regular. There was a prominent left ventricular third heart sound, accentuated pulmonary second sound and a soft systolic murmur in the mitral area. He was also noted to have frontal baldness, drooping eyelids, wasting of the masseters, myotonia, small firm testes, and lenticular opacities on slit lamp examination. There was no significant muscle weakness.

Electrocardiography showed a sinus tachycardia, complete left bundle branch block and left anterior hemiblock. Chest X-ray confirmed left ventricular failure. Echocardiography revealed a moderately dilated left ventricle, hypokinesia of the interventricular septum and left ventricular posterior wall, E point septal separation of 25 mm and a reduced aortic valve opening of 1.2 cm. These parameters were suggestive of moderate to severe left ventricular dysfunction complicating dilated cardiomyopathy.

Dilated cardiomyopathy is rare in adults with myotonia dystrophica. Why this should be so is unknown, as cardiac conduction tissue, which is commonly involved, both sub-clinically and clinically, has a close embryologic link with cardiac muscle. Perhaps the susceptibility of the myocardium to the biochemical defect affecting conduction tissue is determined by hitherto unknown genetic or environmental risk factors. It is also uncommon for patients with myotonic dystrophy to present with severe left ventricular failure as our patient did. Cardiac muscle involvement is commonly occult throughout life and there is no consistent correlation with skeletal muscle involvement.\(^1\) Electrocardiological and electrophysiological studies show abnormal sinus node function in about 20% and His-Purkinje disease in 80% of patients.\(^1\) Isolated left bundle branch block and left anterior hemiblock may occur in 13–24% of patients.\(^1\) But it is rare for a combination of left anterior hemiblock and complete left bundle branch block to occur in the same patient. This may have been a marker of severe cardiac muscle involvement.

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References


Intussusception in ileostomy in a pregnant woman

Sir,

Intussusception in stomata is rare\(^1,2\) and only two cases have been reported previously in the literature reviewed since 1950.\(^3,4\) A case of intussusception of ileostomy in a woman pregnant with twins is presented and its mechanism described.

A 31 year old woman with ileostomy presented with ‘prolapse’ of her stoma, associated with colicky abdominal pain. She was 21 weeks pregnant with twins. Abdominal examination showed a uterus compatible with her date of pregnancy, multiple scars, no area of tenderness and normal bowel sounds. Her haematological investigations were within normal limits.
She had had total colectomy and ileostomy 8 years previously for ulcerative colitis. She had had her ileostomy refashioned 3 times for parastomal hernia, parastomal abscess and retraction.

Several unsuccessful attempts were made to reduce the 'prolapse' of her ileostomy. Examination under anaesthesia revealed proximal ileum intussuscepting through the stoma. The intussusception was about 15 cm from the most distal part of the ileostomy spout. The ileum was slightly congested with no other pathology. Retrograde reduction was unsuccessful and laparotomy was carried out.

At laparotomy it was noticed that there was herniation of one wall of the ileum through a defect in the abdominal wall next to the stoma. This part of the ileum went on to prolapse through the spout and was followed by more ileum to form the intussusception. Reduction was achieved by gentle traction, the lateral space to the stoma was closed and the ileum was attached to the anterior abdominal wall with absorbable sutures. She was managed post-operatively on nitroprusside hydrochloride, a myometrial relaxant.

She went into labour at 35 weeks gestation and had emergency lower segment Caesarian section for fetal distress. Both babies were normal.

A case of intussusception in ileostomy in a pregnant woman has been described by Priest et al. and in a patient with loop colostomy by Keane and Whittaker. In both cases and in the case presented above, there were no aetiological factors and the patients all survived. Diagnosis is easily confused with prolapse, which is easily reducible and for which local revision is usually adequate. Surgical management was different in the 3 cases. In the case described by Priest et al., the patient was managed by revision of her ileostomy and in the case of Keane and Whittaker, by resection and refashioning of the colostomy.

Intussusception as a complication of stomata is not described in textbooks and should be considered if an apparent prolapse proves difficult to reduce or manage locally.

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Gastric leiomyoma: an unusual presentation
Sir,
Leiomyoma is the commonest benign tumour of the stomach found at autopsy. Clinically significant gastric leiomyomas are rare. We report the case of an elderly patient in whom an extraluminal gastric leiomyoma presented with symptoms and signs suggestive of large bowel pathology.

An 84 year old woman presented with a 3 month history of left-sided abdominal pain and altered bowel habit, principally increasing constipation. General examination was normal with, however, suspicion of a palpable upper descending colon. Haematological and biochemical investigations were normal and a barium enema demonstrated only mild diverticular disease of the sigmoid colon. She was commenced on mebeverine and discharged from further review.

She was re-referred 3 months later on account of persistence of the abdominal pain and constipation, accompanied now by a new symptom of anorexia. She had lost 3 kg in weight, and a definite mass was now palpable in the left hypochondrium. Flexible sigmoidoscopy to 50 cm was normal as was a repeat barium enema which again showed only mild sigmoid diverticulosis. Abdominal ultrasound confirmed a left hypochondrial mass (8 cm in diameter), consistent with a tumour arising from either the stomach or pancreas. Both barium meal and upper gastrointestinal endoscopy showed the posterior wall of the stomach to be deformed by extrinsic compression.

An abdominal CT scan demonstrated the mass to be arising from the posterior wall of the stomach. Biopsy under CT guidance yielded only small fragments of tissue from which histology was initially reported as being consistent with a nerve sheath tumour. Laparotomy revealed a large spherical extraluminal tumour (10 x 10 cm across) attached by a narrow pedicle to the posterior gastric wall; histology showed a benign gastric leiomyoma. Two months later she was asymptomatic and had gained over 4 kg in weight.

We could not find a published case similar to ours of an elderly patient with a gastric leiomyoma presenting in such a manner. Anorexia and weight loss are usually early rather than late presenting symptoms, and abdominal pain is more a feature of intestinal than gastric tumours. Our patient's initial palpable 'fullness' must have been the leiomyoma in its early stage, and compression of her left hemicolon most likely explains her altered bowel habit. Although double-contrast barium meal has been the 'gold standard' for many years in the diagnosis of gastric leiomyoma, the value of abdominal CT is now well established, especially in cases of extraluminal extension of tumour.

Gastric leiomyoma is worth considering in the differential diagnosis of unexplained abdominal pain, altered bowel habit and a left hypochondrial mass in an elderly patient, as early diagnosis can lead to a successful clinical outcome.

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