SHORT REPORT

Fibromuscular dysplasia mimicking Crohn's disease over a period of 23 years

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Received 14 July 2011; received in revised form 24 September 2011; accepted 26 September 2011

Abstract

Rare diseases with similar clinical presentation as more frequent gastrointestinal disorders might be challenging in the diagnostic and therapeutic management. In this case we report on a 47-year-old woman who was thought to suffer from Crohn's disease. Symptoms, macroscopic and histological aspects of the gastrointestinal tract, treatment response and clinical course had encouraged the wrong diagnosis over a period of 23 years. After the patient died in the context of a sudden clinical deterioration, fibromuscular dysplasia of the aorta was finally unmasked by post-mortem examination as underlying cause of all symptoms attributed to Crohn's disease. Re-evaluation of former diagnostic procedures revealed subtle aspects of fibromuscular dysplasia, even in biopsy samples from 23 years ago. This first case report of fibromuscular dysplasia of the aorta documents a rare pitfall in the diagnostic workup of a frequent clinical presentation in gastroenterology.

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1. Introduction

Crohn's disease is a chronically relapsing inflammatory condition that may affect the whole gastrointestinal tract. Acute episodes of the disease go together with symptoms like abdominal pain and diarrhoea. Although these episodes can be managed by administration of anti-inflammatory drugs, such as cortisone or other immunosuppressive agents, patients suffering from Crohn's disease are at a high risk to undergo surgery because of local complications like gastrointestinal strictures, abscesses or fistulas. Ileocolonoscopy with biopsies serves as gold standard procedure to establish a reliable diagnosis of Crohn's disease. Macroscopic appearance during endoscopy varies from unspecific signs of inflammation like diffuse redness and mucosal vulnerability to severe mucosal damage including aphthous lesions up to deep ulcerations. Histologically, the disease is characterised by a transmural...
inflammation with deep ulceration, neutrophilic infiltration, granulomas and lymphoid aggregates.\textsuperscript{1,2}

In those cases where only some of these typical criteria are met, diagnosing can be challenging, since other diseases may mimic the clinical picture of Crohn’s disease. Furthermore, response to established therapies may reinforce the wrong diagnosis of Crohn’s disease which can lead to lifelong suffering with potentially fatal outcome as seen in our case reported below.\textsuperscript{3}

2. Case report

We report on a 47-year-old woman who was transferred to our department for clarification of anorexia and abdominal pain in the context of Crohn’s disease, diagnosed 23 years ago.

Back then, when she was hospitalised because of diarrhoea and abdominal pain for the first time, the diagnosis of Crohn’s disease was based on the macroscopic findings during colonoscopy, showing aphthous lesions and fibrin-covered ulcers in the left colon surrounded by normal mucosa, in combination with the histological result of colonic mucosal biopsies, reporting inflammatory infiltration of the mucosa compatible with the clinical diagnosis of Crohn’s disease. Pre-existing iridocyclitis was thought to underline the diagnosis, as this condition is typically associated with Crohn’s disease.

Under short-term treatment with cortisone and continuous therapy with sulfasalazine the disease remained clinically inapparent for eleven years. Then an acute aggravation, accompanied by a stricture of the descending colon, led to sigmoidectomy. The histological assessment of the resected specimen revealed inflammatory changes, compatible with Crohn’s disease of the descending colon with stricture and meso-colonic abscess. During the following years the patient presented with symptoms of acute shock and was transferred to our intensive care unit in comatose state. Acute lab results revealed a haemoglobin level of 5.3 mg/dl, a lactate level of 18 mmol/l and signs of an acute liver failure with massive elevated transaminases (alanine aminotransferase of 11.763 U/l) and low coagulation levels (normotest 10%). Acute gastroscopy showed a grey mucosa from the distal oesophagus to the small intestine with a very vulnerable surface and signs of diffuse mucosal bleeding. Abdominal computer tomography found an intestinal pneumatosis and a diffuse malperfusion of the small intestine, liver and spleen, caused by a complete occlusion of the celiac trunk and the superior mesenteric artery. An acute percutaneous transluminal angioplasty for the purpose of recanalisation was intended but had to be cancelled, since there was no evidence for a remaining perfusion of the occluded vessels on digital subtraction angiography. The patient died with the signs of acute shock.

Autopsy revealed generalised organ ischemia due to fibromuscular dysplasia (FD) of the aorta and consecutive stenosis of all abdominal arteries. The diagnosis of intimal and medial FD was confirmed histologically (Fig. 2).
On the basis of this new information, we re-evaluated all available mucosal biopsies. Thereby we did not find histological features typical for Crohn's disease like transmural or fissural ulcerations or granulomas, for example. Furthermore, we saw histological signs of chronic ischemia like microthrombosis as FD-associated changes (Fig. 3), even in biopsies from the first clinical onset 23 years ago. Additionally, re-evaluation of a CT scan performed 10 days before the patient died detected a partial occlusion of the celiac trunk and a total occlusion of the superior mesenteric artery with collateral feeding from the celiac trunk (Fig. 4).

3. Discussion

Fibromuscular dysplasia (FD) is a non-atherosclerotic, non-inflammatory disease. Histopathologically, FD is characterised by a fibromuscular proliferation of the arterial wall (Fig. 2). Depending on the arterial layer showing the most significant changes, three subtypes of FD can be differentiated. Among those, medial FD is the most frequent form, whereas intimal and adventitial (also known as periarterial) FD are hardly reported. FD mainly affects medium and small arterial vessels like the renal or the internal carotid arteries. Involve-ment of the abdominal arteries is very rare and can be over-looked during the diagnostic work-up of gastrointestinal symptoms.

To our knowledge this is the first case report of fibromuscular dysplasia of the abdominal aorta. In retrospect, the disease was responsible for the gastrointestinal symptoms and other complications during the 23-year-follow up of the patient, including malperfusion of the placenta, 50% stricture of the external carotid arteries and, most typically, renal infarction, probably causing the episode of hypertension. Its gastrointestinal involvement showed features, typically seen in Crohn's disease like aphtous lesions and ulcerations that improved under anti-inflammatory therapy. The clinical progression of the disease was quite similar to the normal fluctuating course of Crohn's disease as well, including surgical interventions due to local complications. The protracted and eventually lethal clinical course of our patient underlines that rare diseases should be kept in mind as they might mimic symptoms of more frequent gastrointestinal disorders.

Conflict of interest

None

Acknowledgements

WD collected all case-related material and drafted the manuscript. JM, FK and FW evaluated examinations that
are discussed in the manuscript and provided discipline-specific background. ChM coordinated the project and helped to draft the manuscript. All authors read and approved the final manuscript.

References