Clin Rheumatol (2005) 24: 76–78
DOI 10.1007/s10067-004-0977-5

CASE REPORT

Erkan Kozanoglu · Sibel Basaran · M. Kamil Goncu

Proximal myopathy as an unusual presenting feature of celiac disease

Received: 20 February 2004 / Accepted: 18 June 2004 / Published online: 2 September 2004

Abstract A 37-year-old woman presented with back pain, diffuse musculoskeletal pain, and muscle weakness without marked gastrointestinal symptoms. She complained of difficulty in walking and bilateral hip pain for the preceding year. Clinical examination revealed proximal muscle weakness especially in the lower extremities and a waddling gait pattern. Laboratory parameters and radiographic findings revealed the diagnosis of osteomalacia. The etiology of osteomalacia was investigated and a diagnosis of celiac disease was established. As osteomalacia symptoms may be the only presenting feature of celiac disease, it should be considered in the differential diagnosis of patients presenting with proximal muscle weakness and diffuse musculoskeletal pain.

Keywords Celiac disease · Looser's zones · Myopathy · Osteomalacia

Introduction

Celiac disease is a complex autoimmune disease which is characterized by a strong genetic association (HLA-DQ2 or DQ8) [1]. It is an inflammatory disorder of the small intestine and clinical manifestations range from asymptomatic to severe malabsorption [2, 3]. In adults common presentations are anemia and variable abdominal symptoms. Less commonly, patients present with a more typical history with features of steatorrhea, weight loss, bruising, and other symptoms of nutritional deficiencies resulting from malabsorption [2]. Patients with overt celiac disease will occasionally have evidence of osteomalacia [4]. However, it is rare for celiac disease to present only with osteomalacia symptoms such as proximal muscle weakness, diffuse musculoskeletal pain, and fractures in the absence of a specific patterns [4, 5, 6, 7, 8, 9, 10, 11]. Herein we present a patient with osteomalacia symptoms who had previously undiagnosed celiac disease.

E. Kozanoglu (✉) · S. Basaran · M. K. Goncu
Department of Physical Medicine and Rehabilitation,
Cukurova University Medical Faculty,
01330 Adana, Turkey
E-mail: ekozanoglu@yahoo.com
Tel.: +90-332-3386429
Fax: +90-332-3386429

Case report

A 37-year-old woman presented with a 6-year history of back pain, progressively increasing diffuse musculoskeletal pain, and muscle weakness. She also complained of fatigue, tiredness, bilateral hip pain, difficulties in rising from a chair, holding her arms up, walking, and inability to ascend stairs for the preceding year. She had mild diarrhea for 8 years, which was considered as irritable bowel syndrome and treated symptomatically. She also had a history of weight loss in this period.

On physical examination; her height was 152 cm and weight was 47 kg. She was pale and no other significant finding was detected. Neuromuscular examination revealed proximal muscle weakness, especially in the lower extremities, and hypoactive reflexes in four extremities. Hip flexor and abductor strength was 3/5 bilaterally. Hip range of motion was limited and painful. She had a waddling gait pattern. She also experienced pain on her spine by percussion.

Laboratory investigations showed iron deficiency anemia [hemoglobin: 7.96 g/dl (12–16 g/dl)]. Elevated serum alkaline phosphatase (ALP) of 1488 U/l (50–305 U/l), decreased calcium of 7.7 mg/dl (8–10.6 mg/dl), and inorganic phosphorus of 1.6 mg/dl (2.5–4.5 mg/dl) were found. The 25-hydroxy vitamin D level was 11.5 ng/ml (14–75 ng/ml) and parathyroid hormone level was 268 pg/ml (8–65 pg/ml). Other laboratory investigations including erythrocyte sedimentation rate (ESR), C-reactive protein (CRP), thyroid function tests, and creatinine kinase were normal. Antinuclear antibodies (ANA) and anti-DNA were negative. Parathy-

roid scintigraphy showed no pathological changes. Radiographs of the thoracic and lumbosacral spine demonstrated osteoporotic vertebral collapse, prominence of vertical trabeculation, and increased thoracic kyphosis. Bilateral fracture lines on the pubic rami and Looser's zones (pseudofractures) on both femurs, which are characteristic for osteomalacia, were observed on pelvic and femur X-rays (Fig. 1). Looser's zones were also seen on the bilateral tibia. Osteoporosis was confirmed by the dual energy X-ray absorptiometry (DEXA) method. The T score for the lumbar spine was −6.05 and for the femoral neck was −2.10.

As the patient had anemia, mild diarrhea, weight loss, and osteomalacia, investigations for malabsorption were carried out and antigliadin IgA and IgG antibodies were found to be elevated [IgA: 26.2 (0–12), IgG: 92.2 (0–12)]. Subsequent duodenal biopsy confirmed a diagnosis of celiac disease. She was started on a gluten-free diet and also given vitamin D, calcium, and iron supplementation.

Within 3 months her complaints including muscle weakness and diffuse musculoskeletal pain resolved. She gained 3 kg in 3 months. Laboratory parameters were also improved within 6 months (25-hydroxy vitamin D: 48 ng/ml, parathyroid hormone: 110 pg/ml, ALP: 223 U/l, Ca: 8.4 mg/dl, inorganic phosphorus: 3.7 mg/dl, antigliadin IgA: 20.9, and IgG: 19.2). Bone mineral density had increased markedly after 1 year and the T scores for the lumbar spine and femoral neck were −0.99 and 0.78, respectively. Duodenal biopsy could not be repeated as the patient did not accept the biopsy procedure.

Discussion

Common presentation of celiac disease includes malaise, lethargy, diarrhea, abdominal pain, and weight loss. Tiredness and anemia are frequently the only presenting features of celiac disease [12]. However, presentation with osteomalacia, myopathy, peripheral neuropathy, and malnutrition is less common [4]. In adults, celiac disease may be subclinical and difficult to diagnose. Hin et al. [12] reported that many celiac patients present with nongastrointestinal symptoms especially with anemia. Our patient presented with diffuse musculoskeletal pain and proximal muscle weakness.

It was first described in 1965 that osteomalacia can occur without malabsorption symptoms [5]. Later it was reported that osteomalacia could be the only presenting symptom in celiac disease [6]. Several case reports of celiac disease presenting with bone pain, proximal myopathy, and characteristic radiographic findings have been reported in the literature to date and most of them were from northern latitudes [6, 7, 10, 13, 14]. Osteomalacia due to celiac disease is less frequently reported from sunnier climates [8, 9, 15]. Adana is a city located in the south of Turkey near the Mediterranean Sea with a sunny climate more than half of the year. We think that vitamin D deficiency and osteomalacia caused by malabsorption remained asymptomatic for a long time in our patient because of the compensation of endogenous vitamin D production by excessive sun exposure.

Secondary hyperparathyroidism develops as a consequence of reduced vitamin D and calcium levels due to malabsorption and causes increased bone turnover. Reduced physical activity due to widespread pain in combination with hypocalcemia and secondary hyperparathyroidism results in low bone mineral density [12, 16]. Radiological evidence of osteoporosis in the form of vertebral collapse is frequently observed, but low impact fractures of the hip or pubic rami are rarely seen in celiac disease as in our patient and this should alert the clinician to the possibility of osteomalacia [7]. In our patient, osteoporosis of both proximal femurs together with fractures on pubic rami is the main cause of bilateral hip pain and limitation of range of motion. For the treatment of bone disease, vitamin D and calcium supplementation with or without bisphosphonates is recommended [2, 7]. In a case report, Byrne et al. [4]

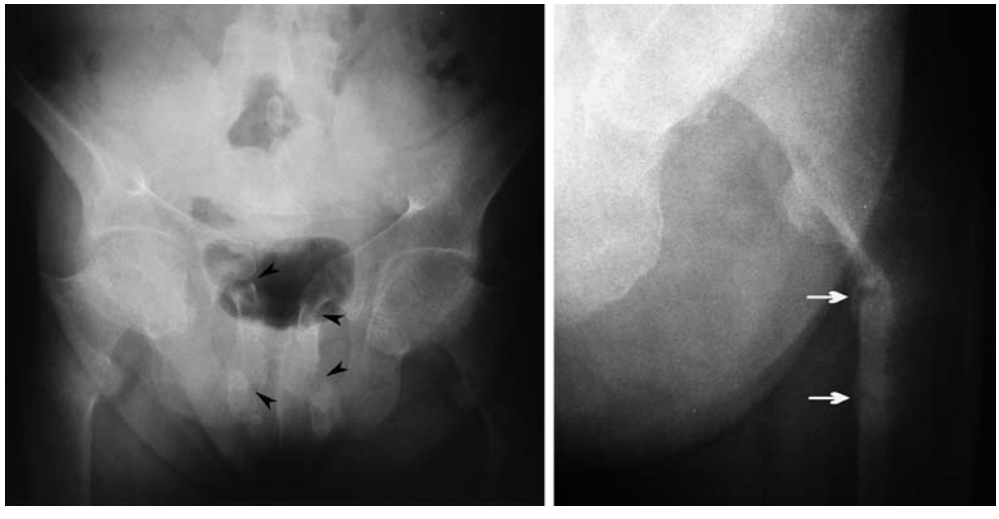

Fig. 1 Bilateral fracture lines on pubic rami are shown by *arrowheads* (left side) and Looser's zones on left femur are shown by *arrows* (right side)

demonstrated that treatment with vitamin D, calcium, and gluten-free diet caused resolution of pain, restoration of normal muscle strength, and resulted in weight gain in 3 months. Our patient was also completely symptom free after 3 months. Although she had severe osteoporosis, bone mineral density was normalized within 1 year.

The endomysial antibody has proved to be the most useful test with a specificity and sensitivity of 90–95% especially in younger patients [2, 17]. However, Byrne et al. [4] found antiendomysial antibody to be negative in their case report. We observed higher levels of antigliadin antibodies in our patient, but unfortunately we could not investigate the antiendomysial antibody.

Celiac disease is frequently underdiagnosed or misdiagnosed and results in unnecessary morbidity such as disabling osteomalacia and fractures [12]. Therefore, early diagnosis and treatment of celiac disease is important because the symptoms completely resolve with adequate treatment.

Osteomalacia-associated myopathy may be the only presenting feature of celiac disease and it should be considered in the differential diagnosis of patients presenting with proximal muscle weakness and diffuse skeletal pain with accompanying anemia.

References

1. Dieterich W, Esslinger B, Schuppan D (2003) Pathomechanisms in celiac disease (abstract). Int Arch Allergy Immunol 132:98–108
2. Jewell DP (2003) Coeliac disease. In: Warrell DA, Cox TM, Firth JD, Benz EJ (eds) Oxford Textbook of Medicine. Oxford University Press, New York, pp 585–589
3. Schuppan D (2000) Current concepts of celiac disease pathogenesis. Gastroenterology 119:234–242
4. Byrne MF, Razak AR, Leader MB, Sheehan KM, Patchett SE (2002) Disabling osteomalacic myopathy as the only presenting feature of coeliac disease. Eur J Gastroenterol Hepatol 14:1271–1274
5. Moss AJ, Waterhouse C, Terry R (1965) Gluten-sensitive enteropathy with osteomalacia but without steatorrhea. N Engl J Med 272:825–830
6. De Boer WA, Tytgat GN (1992) A patient with osteomalacia as a single presenting symptom of gluten sensitive enteropathy. J Intern Med 232:81–85
7. Basu RA, Elmer K, Babu A, Kelly CA (2000) Coeliac disease can still present with osteomalacia. Rheumatology 39:335–336
8. Jawad AS, Squire SB, Perry JD (1988) Osteomalacia and coeliac disease presenting as isolated dactylitis. Ann Rheum Dis 47:692–694
9. Hajjar ET, Vincenti F, Salti IS (1974) Gluten induced enteropathy: osteomalacia as its principal manifestation. Arch Intern Med 134:565–566
10. Wong M, Scally J, Watson K, Best J (2002) Proximal myopathy and bone pain as the presenting features of coeliac disease. Ann Rheum Dis 61:87
11. Garcia-Porrua C, Gonzales-Gay MA, Avila-Alvarenga S, Rivas MJ, Soilan J, Penedo M (2000) Coeliac disease and osteomalacia: an association still present in Western countries. Rheumatology 39:1435
12. Hin H, Bird G, Fisher P, Mahy N, Jewell D (1999) Coeliac disease in primary care: case finding study. BMJ 318:164–167
13. Hardoff D, Sharf B, Berger A (1980) Myopathy as a presentation of coeliac disease (abstract). Dev Med Child Neurol 22:781–783
14. Cimaz R, Bazzi P, Prelle A (2000) Myopathy associated with rickets and celiac disease. Acta Paediatr 89:496–497
15. Jain V, Angitii RR, Singh S, Thape BR, Kumar L (2002) Proximal muscle weakness—an unusual presentation of coeliac disease. J Trop Pediatr 48:380–381
16. McFarlane XA, Bhalla AK, Robertson DAF (1996) Effect of a gluten free diet on osteopenia in adults with newly diagnosed coeliac disease. Gut 39:180–184
17. Unsworth DJ (1996) Serological diagnosis of gluten sensitive enteropathy. J Clin Pathol 49:704–707