Celiac disease, known as celiac sprue is a gluten-sensitive enteropathy characterized by malabsorption resulting from inflammatory injury to the mucosa of the small intestine. It is well known to be associated with a variety of neurological disorders including epilepsy, myopathy, neuropathy and ataxia. The nature of this association is unclear. Although osteomalacia secondary to vitamin D deficiency is a recognized complication of celiac disease, however, severe osteomalacic myopathy as the only presentation of celiac disease is extremely rare. We present 2 interesting cases of osteomalacic myopathy secondary to celiac disease, which were treated successfully with full recovery. An important and unique observation was the brisk reflexes noticed in both patients. The mechanism behind this phenomenon is not well understood. Work-up for celiac disease is warranted in any young patient that presents with myopathy.

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Patient 1.  A 43-year-old Saudi woman presented with progressive weakness of her legs, which started 8 months prior to presentation. The weakness was noticed when walking for a long distance or climbing stairs. It involved the upper limbs too, but to a lesser extent. It progressed to the point that she was not able to walk. The weakness was not fluctuating, no muscular pain or cramps. No sensory symptoms, speech or swallowing difficulties, cranial nerve involvement, skin rash, seizures, headache, fever, sphincter involvement or joint pain. Systemic review was unremarkable. Past history was unremarkable as well as her social and family history. She is married with 4 children. Her physical examination showed normal higher mental function. Cranial nerve examination was normal. Motor examination showed normal tone. She had a generalized weakness, proximal (3+/5) more than distal (4/5). Reflexes were brisk all over (3), marked in knees jerks (+3) with down-going toes. Sensory examination was normal. Coordination was normal. She had a waddling gait with positive Gower's sign. Her investigations showed low hemoglobin of 8.3 gm/dl with microcytic hypochromic picture, normal white blood cells and platelets. Renal profile was normal. Calcium level was low at 2.13 mmol/L (2.15-2.55), as well as phosphates 0.77 mmol/L (0.87-1.45). Liver function tests showed high alkaline phosphatase (ALP) 739 IU otherwise unremarkable. Thyroid function test, erythrocyte sedimentation ratio and connective tissue screen were normal. Vitamin D level was very low (<12 nmol/L). She had an elevated anti-gliadin A antibody (AGAA) level at 42.4 (0-10 IU/ml) with normal acid glycosaminoglycans (AGAG). Endomysial antibody (ENDMA) was not carried out, HIV, syphilis, and brucella serologies were negative. Humerus x-ray was normal. Femur and pelvis x-rays showed Looser's zone, diffuse osteoporosis, and hip fracture at the inferior aspect of right femoral neck (Figure 1). Lumbosacral spine and sacroiliac joint x-ray showed mild osteoporosis; otherwise, normal. A CT bone mineral densitometry is 2 SD below the mean for the age-matched normal. However, it remains above the fracture threshold. Nuclear medicine bone scan showed homogenous tracer uptake at the skeleton with multiple focal increased tracer activity at the ribs on sides, sternum, cervical spine and right ischium (Figure 2). These most likely represent fractures. Upper gastrointestinal tract endoscopy with gastric and jejunal biopsy was carried out. Biopsy showed villous atrophy with moderate lymphocytic infiltration. Skin biopsy showed atopic dermatitis. She had normal nerve conduction studies. An EMG showed classical small polyphasic waves suggestive of myopathic process. Magnetic resonance imaging of both brain and cervical spine was unremarkable. The patient was started on a gluten free diet, vitamin D, calcium and iron supplements. She was advised to take a lot of dairy product and expose her skin to the sun. Her power and gait were normal 8 weeks later during an outpatient follow-up visit.

Patient 2. A 32-year-old Saudi woman presented with progressive weakness of her legs, which started 8 months prior to presentation. The weakness was noticed when walking for a long distance or climbing stairs. It involved the upper limbs too, but to a lesser extent. It progressed to the point that she was not able to walk. The weakness was not fluctuating, no muscular pain or cramps. No sensory symptoms, speech or swallowing difficulties, cranial nerve involvement, skin rash, seizures, headache, fever, sphincter involvement or joint pain. Systemic review was unremarkable. Past history was unremarkable as well as her social and family history. She is married with 4 children. Her physical examination showed normal vital signs. Apart from being overweight, her ENT, chest, cardiovascular system and abdomen were normal. Neurological examination showed normal higher mental functions, cranial nerves II to XII, tone and sensations. There is no atrophy, fasciculation or tenderness. She was globally weak, proximal more than distal, legs are weaker than arms. Reflexes were brisk all over. Planter responses were downgoing, coordination was normal. She had a waddling gait. Investigations showed hemoglobin of 11.9, microcytic hypochromic picture. Urea and electrolytes normal, iron low, ALP 148, alanine aminotransferase normal, phosphates 0.7, calcium 1.2, vitamin D <12. Thyroid function test normal, creatine kinase normal, AGAA 18 (0-11), AGAG

Figure 1 - Femur and pelvis x-rays shows Looser's zone in the medical cortex of the right femoral neck, diffuse osteoporosis, and hip fracture at the inferior aspect of pubic ramus.

Figure 2 - Nuclear medicine bone scan showed homogenous tracer uptake at the skeleton with multiple focal increased tracer activity at the ribs on sides, sternum, cervical spine and right ischium.
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26.5 (0-11). ENDMA 39. Femur and pelvic x-ray, magnetic resonance imaging of both brain and cervical spine was unremarkable. Upper gastrointestinal endoscopy with gastric and jejunal biopsy was carried out. Biopsy showed villous atrophy with moderate lymphocytic infiltration (Figures 3a & 3b). She was treated with the same regimen (as the first patient) and improved gradually. She is currently, able to walk, climb up-stairs and perform activities of daily life and self-hygiene.

Discussion. Celiac disease is well known to be associated with a variety of neurological disorders including epilepsy, myopathy, neuropathy and ataxia. The nature of this association is unclear and different mechanisms have been proposed including vitamins and trace element deficiencies, altered autoimmunity, heredity, and gluten toxicity. Muscle diseases associated with celiac disease include myopathy, polymyositis, dermatomyositis, myotonic dystrophy, muscular dystrophy and rhabdomyolysis. Myopathy as the only presentation of celiac disease is extremely rare. Usually, the patient will have other gastrointestinal or systemic involvement (including nervous system). In our case, myopathy was the only presenting feature. Another important and unique observation was the brisk reflexes noticed in both patients. They were so marked to the degree that a workup for myelopathy was carried out. The mechanism behind this phenomenon is not well understood. We think that a work-up for celiac disease is warranted in any young patient presenting with myopathy.

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