Late Diagnosis Of Celiac Disease In An Adolescent Girl With Multiple Fractures : A Case Report

AbdulRahman A. Sari, AbdulRahman A. Hummadi

Abstract— 14-year-old female patient was referred to endocrine clinic in Jizan, Saudi Arabia, in January 2015 as a case of osteogenesis imperfecta with history of multiple fracture, started at the age of 8 years after minimal trauma with progressive muscle weakness, patient know on wheel chair. She was diagnosed with celiac disease based upon serologic results and the biopsy findings, a gluten free diet start, and the patient improve dramatically.

Index Terms— Celiac disease, Multiple fractures, osteogenesis imperfecta, Case Report; Saudi Arabia.

INTRODUCTION:

Celiac disease is a serious Autoimmune disorder in which the mucosal lining of the small intestine is damaged in response to ingestion of gluten and similar proteins, which are found in wheat, oats, rye, barley and other grains[1]. Celiac disease is a common intestinal disorder that is often confused with other conditions. It causes severe intestinal damage manifested by several uncomfortable signs and symptoms[2,3].

CASE REPORT:

14-year-old female patient was referred to endocrine clinic in Jizan, Saudi Arabia in January 2015 as a case of osteogenesis imperfecta with history of multiple Fracture started at the age of 8 years after minimal trauma with progressive muscle weakness, patient know on wheel chair. No history of Abdominal distension or diarrhea.

On physical examination, the patient communicated well, alert, conscious . Her height was 120 cm, she weighed 29 kg and her body mass index was 20.14 kg/m2. Patient on wheel chair, her Power 3/5 with normal reflex and tone. Breast development with pubic hair growth was categorized as Tanner stage I. Further physical examination of Chest, CVS and Abdomen is no abnormality detected.

Laboratory findings revealed Complete blood count showed the following values: Hb: (8.4 g/dL), Hct: (29.4%), WBC: (4.92 X10^9/L), Plt: (493 X10^9/L), MCV: (59.6 Fl), MCH: (17 pg), MCHC: (28.6 g/dl), RDW (28.8%). The serum ferritin is low: (7.2 ng/L) .The routine biochemical tests showed Sodium (142 mmol/L, normal: 135-145), Potassium (4.42 mmol/L, normal: 3.5-5.5), low calcium (1.97 mmol/l, normal: 2.2-2.62), and magnesium (0.89 mmol/l, normal: 0.74-1), total Protein (74 g/L, normal: 64-82), Albumin (41.9 g/L, normal: 34-50), and elevated levels of alkaline phosphatase (536 U/L, normal: 50-137). The other biochemical results were within normal limits and routine urine examination was normal. Thyroid function tests showed Free T3 level of 6.9 PMOL/L slightly increased (normal: 3.1-6.8), normal Free T4 level of 12 pmol/L (normal: 12-22) and normal TSH concentration of 1.3 µIU/mL (normal: 0.30-4.00). The concentrations of PTH (14.18 pmol/L, normal: 1.6-6.9), and VIT. B12: (99.6 pmol/L, normal:156-698) and low serum 25-hydroxyvitamin D (6 ng/mL, normal: 10-40) were also measured. The results of other hormonal assay were within normal limits. The serological tests for various infectious agents (HBsAg, anti HBC, Brucella Ig G/M antibody levels, TORCH titers) were found to be negative. She was found to have positive Anti Tissue transglutaminase (Anti TTG). Positive TTG IgG antibody 54 IU/ml and TTG IgA antibody (137 IU/ml) levels were also detected.

Duodenal biopsies was done and histopathology revealed complete villous atrophy, increase in lamina propria inflammatory cell contents including lymphocytes, plasma cells and few scattered neutrophils along with increase in intraepithelial lymphocytes (at rate of >30/100 epithelial cells). No evidence of granulomas or microorganism seen. No evidence of dysplasia or malignancy in the examined tissue [Figure1].

Final diagnosis was celiac disease based on above finding and the patient was managed with gluten- free diet, vitamin and minerals. The patient improve dramatically. Power improve to 5/5 with improve body build, height increase by 8 Cm/year. The patient now can walk with doing daily activity.

DISCUSSION:

The diagnosis of Celiac disease can be established by the combination of a history and physical examination and a diagnostic tests (serologic results and the biopsy findings)[4]. But In our case, the patient was diagnosed with CD based upon serological and histological findings which not compatible with

clinical and laboratory manifestations and noticeable clinical improvement was observed on a gluten-free diet[5].

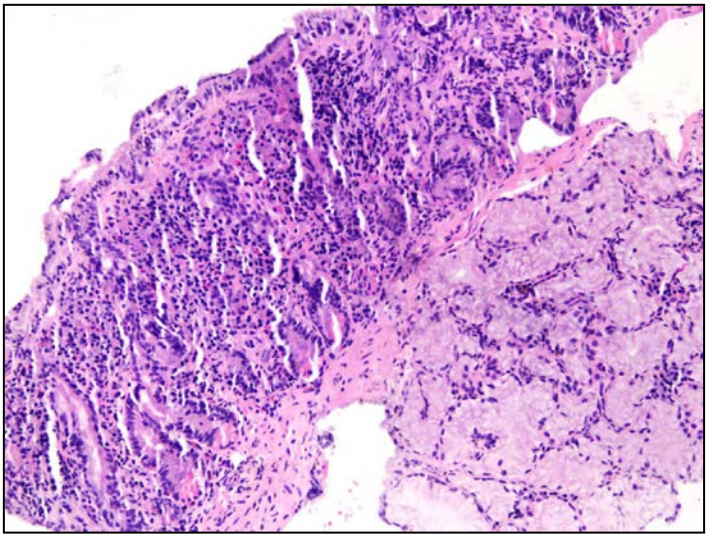
CONCLUSION:

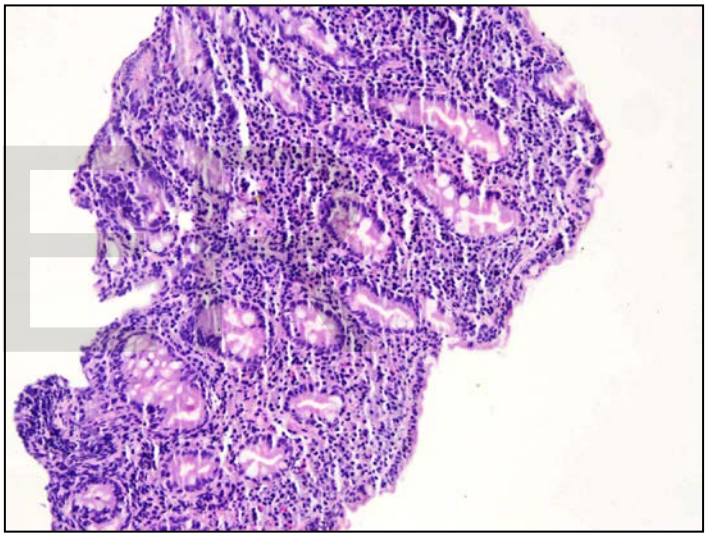
Celiac disease should be suspected and roll out in each patient with some clinical fracture even no GI symptoms, Early recognition of this disease is important to avoid further complication to the patient. Serology and Duodenal biopsies should be performed to confirm the diagnosis.

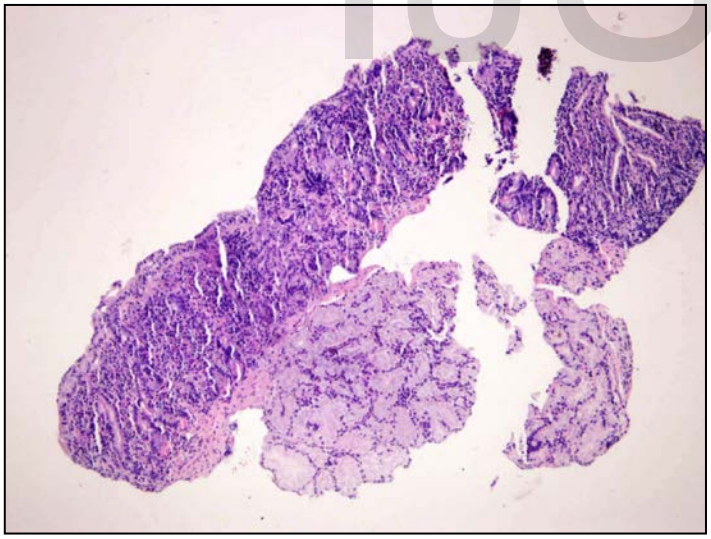
FIGURES:

.

Figure 1,2&3: Photos of Duodenal biopsies shows fragments of duodenal tissue with totally blunted villi, increase in lamina propria inflammatory cell contents including lymphocytes, plasma cells and few scattered neutrophils along with increase in intraepithelial lymphocytes (at rate of >30/100 epithelial cells). No evidence of granulomas or microorganism seen. No evidence of dysplasia or malignancy in the examined tissue.







REFERENCES:

[1] Corazza GR, Gasbarrini G. Celiac disease in adults. Bailliere's Clinical Gastroenterol 1995; 9: 329-50.
[2] https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1323300
[3] https://emedicine.medscape.com/article/171805-overview#a5
[4] Freeman HJ: Refractory celiac disease and sprue-like intestinal disease. World J Gastroenterol. 2008, 14 (6): 828-830. 10.3748/wjg.14.828.
[5] Loftus C, Murray J. Celiac disease: diagnosis and management. J Clin Outcomes Manage. 2002;9:341–350.