VOLUME-9, ISSUE-2, FEBRUARY-2020 • PRINT ISSN No. 2277 - 8160 • DOI : 10.36106/gjra

Original Research Paper

Paediatrics

A RARE CASE OF CELIAC DISEASE PRESENTING AS RICKETS -A CASE REPORT

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ABSTRACT

BACKGROUND: Celiac disease is an immune-mediated disorder that primarily affects the gastrointestinal system and has varied intestinal and extraintestinal manifestation.

CASE CHARACTERISTICS: A 4yr old female child with deformities in the extremities and walding gait since 8 months with failure to thrive.

OBSERVATION: Celiac disease has intestinal manifestations as well as extraintestinal manifestation, In the form of rickets; short stature; peripheral neuropathy, refractory anemia.

MESSAGE: Any case of rickets not responding to vitamin-D supplements should be evaluated for celiac disease to prevent the long term complications.

KEYWORDS : Human Leukocyte Antigen, Tissue Transglutaminase, Endomysial Antibody

INTRODUCTION:

Celiac disease is an autoimmune-mediated condition involving the proximal small intestine and characterized by the abnormal intestinal mucosa and specific serological and histological profile triggered by gluten ingestion in genetically predisposed individuals[1]

It affects 0.6 to 1% of the population. In India, it is more common in the north Indian population and has an incidence of 1%.In the south Indian population, the prevalence of the celiac disease is very less as associated HLA class (HLA DQ2 & DQ8) is three times more common in the north Indian population compare to the south Indian population.

CASE REPORT:

A 4year old female child born through nonconsanguineous marriage was brought with a complaint of deformities of the extremities and walding gait since 8months.there was no h/o abdominal pain or diarrhea.

She was taken to another hospital for the above complaint and diagnosed as rickets and was given vitamin-D supplements, but there was no improvement. The child also has failure to thrive.

O/e: weight 7.8kg (<-3Z score) height 77cms (<-3Z score) pectus carinatum+, rachitic rosary+, Harison sulcus+.

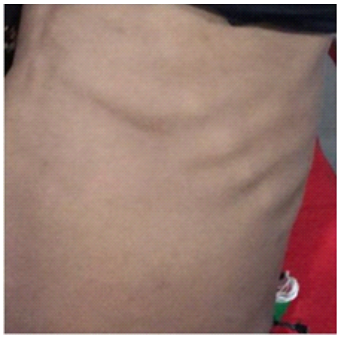

FIG 1-SHOWING HARRISON SULCUS

With all the features of rickets along with failure to thrive, we came to a provisional diagnosis of malabsorption syndrome (?celiac disease).

The child was investigated further. The investigations shows serum calcium9.8mg/dl, serum phosphorus 3.2mg/dl., alkaline phosphatase raised, vit D levels decreased.

X-RAY WRIST showed a widening of growth plates, fraying, and cupping.

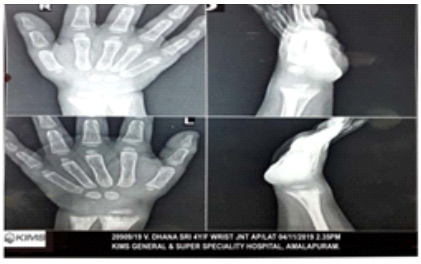

Figure 2-XRAY WRIST JOINT

Serological test are positive for anti tTG and anti-EMA.

Endoscopy of small intestine showed scalloping and fissuring of duodenal folds, which is a consistent finding of celiac disease.

Histopathological report suggestive of severe villous atrophy with increased number of intraepithelial lymphocytes and elongated crypts. According to the marsh- oberhuber grading belongs to grade 3C.

TREATMENT:

The child was started on GLUTEN free diet along with vitamin D and iron supplements.

The child was brought for follow up at 5months after starting treatment and has attained good catch-up growth.

DISCUSSION

Celiac disease or gluten induced enteropathy is a chronic

illness caused by an inflammatory response to gluten proteins[1,2]. The global prevalence is 1% with female to male ratio of 2:1 to 3:1[1,2,3].

In genetically susceptible individuals on dietary exposure to gluten, develop autoimmune reactions, which results in histopathological changes in the small intestine leading to malabsorption[1,3,4].

There still remain many undiagnosed cases of celiac disease. It has shown that for one diagnosed case of celiac disease, there are seven undiagnosed cases in the community[2,3,6].

Classical presentation with chronic diarrhea, failure to thrive, and anemia have become rare. Many patients now present with nongastrointestinal symptoms such as refractory anemia, rickets, short stature, peripheral neuropathy, constipation with abdominal distention, recurrent aphthous stomatitis and dermatitis herpetiformis[1,2,3,4,6,7].

In children with growth retardation and short stature can raise the suspicion of celiac disease.

Measurement of IgA tissue transglutaminase antibody is the preferred initial screening test. The gold standard for celiac disease diagnosis is represented by a combination of mucosal changes detected by duodenal biopsy and by positive serological test i.e.Anti Ttg, Anti EMA, deamidated gliadin peptide[1,3,5,6].

In the recent modification of ESPGHAN criteria, it has proposed that intestinal biopsy is not essential to diagnose celiac disease in a subset of symptomatic patients who have Ttg more than ten times upper limit of normal range along with HLA DQ 2/8 AND EMA positivity[4,6,7].

In India, to diagnose celiac disease, positive serology along with villous atrophy and disappearance of serology with subsidence of symptoms on gluten free diet would make diagnosis of celiac disease.

Treatment: The mainstay of treatment is gluten free diet. Response to gluten free diet is dramatic with diarrhea passive by one week, catch up growth in 6months, and catch height in 12months[4, 6, 7]. A drug larazotide acetate (a tight junction regulator peptide that restores tight junctions integrity and prevents paracellular transport of macromolecules like gluten) been developed and is in phase III trail[5,6,7].

In conclusion, any case of rickets not responding to vitamin D supplements along with features of malabsorption should be evaluated for celiac disease as the response to therapy is excellent so can prevent long term complications.

REFERENCES:

1. Celiac disease: a comprehensive current review G Caio, U Volta, A Sapone, DA Leffler… - BMC …, 2019 - bmcmedicine.biomedcentral.com
2. Celiac disease: From pathophysiology to treatment I Parzanese, D Qehajaj, F Patrinicola… - World journal of …, 2017 - ncbi.nlm.nih.gov
3. Celiac disease: pathophysiology, clinical manifestations, and associated autoimmune conditions JM Barker, E Liu - Advances in pediatrics, 2008 - advancesinpediatrics.com
4. Gluten-Free Diet in Celiac Disease—Forever and for All? A Itzlinger, F Branchi, L Elli, M Schumann - Nutrients, 2018 - mdpi.com
5. Recent advances in celiac disease and refractory celiac disease G Malamut, S Cording, N Cerf-Bensussan - F1000Research, 2019 - ncbi.nlm.nih.gov
6. Piyush guptha textbook of pediatrics
7. Nelson textbook of pediatrics.