



Journal Homepage: -www.journalijar.com

INTERNATIONAL JOURNAL OF ADVANCED RESEARCH (IJAR)

Article DOI:10.21474/IJAR01/20091
DOI URL: <http://dx.doi.org/10.21474/IJAR01/20091>



RESEARCH ARTICLE

CELIAC DISEASE AND DOWN SYNDROME

Parveen Malhotra, Yogesh Sanwariya, Senti, Vishavjeet, Harman Singh and Sandeep Kumar

Department of Medical Gastroenterology, PGIMS, Rohtak, Haryana, India.

Manuscript Info

Manuscript History

Received: 17 October 2024

Final Accepted: 19 November 2024

Published: December 2024

Key words:-

Celiac Disease, Endoscopy, Down Syndrome, Gluten, Gliadin, Prolamins

Abstract

Celiac disease is immune related disorder of small bowel which is seen in genetically predisposed people and issue to permanent intolerance to wheat gliadins and other cereal prolamins also known as gluten-sensitive enteropathy or non-tropical sprue. In 1888, it was first described by Dr. Samuel Gee and in Greek means koiliakos-abdominal. Dicke in 1950 highlighted association between the consumption of bread, cereals and diarrhea which improved after stoppage of wheat intake. The diagnosed cases of celiac disease represent just a tip of iceberg and rest 90% are hidden and undiagnosed. Celiac disease is confirmed by clinical symptoms, serology, endoscopy, histopathological diagnosis and resolution of symptoms after gluten restricted diet. Celiac disease requires life-long gluten restriction; thus, compliance rate varies between 50% to 90%, and is slightly more in elderly & females. As per available data, there is association between coeliac disease and Down's syndrome. Therefore, while evaluating associations of Down's Syndrome, coeliac disease should not be missed.

Copyright, IJAR, 2024, All rights reserved.

Introduction:-

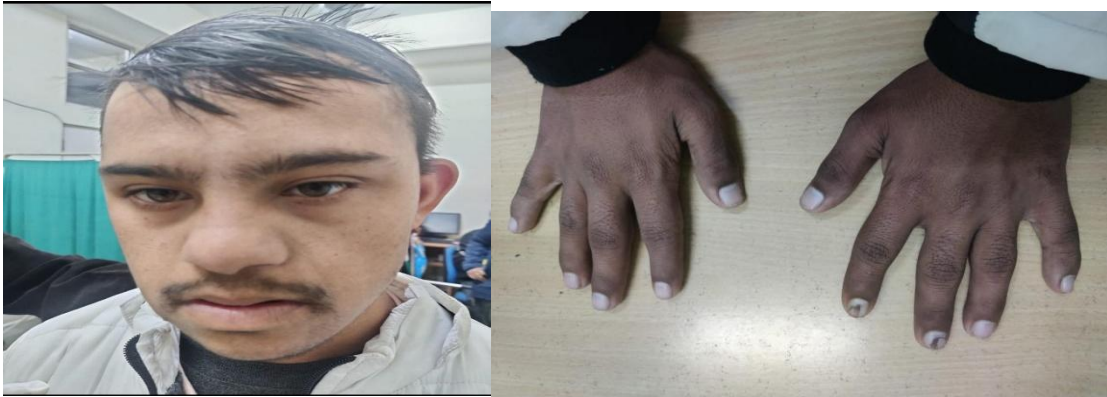
Celiac Disease (CD) is a immune-mediated disorder of small bowel that is seen in genetically predisposed people [1]. Wheat, rye, oat and barley prolamins are the major culprit due to presence of glutamine and proline content in them in significant amount [2]. In the past CD presented in majority of patients with typical gastrointestinal (GI) symptoms but now half of cases present with variety of atypical symptoms or even without any symptoms [3]. Marsh classification is used for histologic changes and vary from presence of intraepithelial lymphocytes to severe villous atrophy. The anti-tissue transglutaminase antibodies are the most sensitive test for CD [2]. The various etiological factors considered for CD include genetic (HLA class II antigen), environmental risk factors [4] and GI infections [5]. The transglutaminase auto antibodies play a role in disease pathogenesis [6]. The prevalence of CD worldwide and in India is globally 1% [7]. Initially, association between Down's syndrome and coeliac disease was very less reported [8-15] but with passage of time more and more cases of their association are being reported [16-23]. The explanation for association is that there are common pathogenetic factors in these diseases, such as histocompatibility (HLA) antigens (HLA-DQ2) [17,19,20,24,25] which are responsible for immune response. The prevalence rates of coeliac disease in patients with Down's syndrome, as reported in literature varies from 2.5 to 18.6% [16-23]

Corresponding Author:- Parveen Malhotra

Address:- 128/19, Civil Hospital Road, Rohtak, Haryana, India (124001).

Case Report

A fifteen-year male who was diagnosed to be suffering from Down's syndrome at seven years of age, presented with long duration of constipation and pain abdomen, for last four years. On evaluation, he was of short stature, flat base and nasal bridge, short and broad hands & foot, intellectual disability with mild anemia. He was able to play outdoor games, on mobile phone but was not able to attend normal school. His cardiological, respiratory, neurological examination was essentially normal. His serum IgA tTG antibodies were massively raised to 180 I.U./ml (normal being 0- 20 I.U./ml) and endoscopy showed severe scalloping of duodenal folds in second part of duodenum and on histopathological examination, Marsh grade 2 celiac disease was diagnosed. He was advised gluten restricted diet which he rigorously followed, thus symptomatic recovery started and he had complete resolution of symptoms within four months, along with increase of hemoglobin to 12 gm% and constipation subsided completely.



Picture 1: - Showing Flat Nose and Face. **Picture 2:** - Showing short thick Fingers.



Picture 3: - Showing short and thick Toes.

Discussion:-

There is definite association between coeliac disease and Down's syndrome, as reported in various studies [16-23]. Thus, whenever any new coeliac disease patient is diagnosed the association with Down's syndrome should always be looked for. There is a high prevalence of immune-related disorders in patients with Down's syndrome, especially autoimmune thyroid disease, diabetes mellitus type I, autoimmune chronic active hepatitis, alopecia, vitiligo, juvenile rheumatoid arthritis or sarcoidosis, [22,26-29]. The Common link between them is immunogenetic markers, particularly HLA antigens [17,24,25,30].

Conclusion:-

Celiac disease and Down syndrome have many independent associations as well as with each other. Hence treating health professionals should be vigilant for timely diagnosis of these cases, as they have many atypical presentations, justifying tip of iceberg phenomenon of these diseases.

Conflict of Interest-

No conflict of interest and prior permission from patient and relatives was taken before publishing the case report.

References:-

1. Harris LA., et al. "Celiac disease: clinical, endoscopic and histopathologic review". *Gastrointestinal Endoscopy* 76 (2012): 625-640
2. Volta U and Villanacci V. "Celiac disease: diagnostic criteria in progress". *Cellular and Molecular Immunology* 8 (2011): 96-102.
3. Serra S and Jani PA. "An approach to duodenal biopsies". *Journal of Clinical Pathology* 59 (2006): 1133-1150.
4. Akobeng AK., et al. "Effect of breast feeding on risk of celiac disease: a systematic review and meta-analysis of observational studies". *Archives of Disease in Childhood* 91 (2006): 39-43.
5. Myleus A., et al. "Celiac disease revealed in 3% of Swedish 12-year-olds born during an epidemic". *Journal of Pediatric Gastroenterology and Nutrition* 49 (2009): 170-176.
6. Guandalini S. "The influence of gluten: weaning recommendations for healthy children and children at risk for celiac disease". *Nestlé Nutrition Institute Workshop Series: Pediatric Program* 60 (2007): 139-151.
7. Plot L and Amital H. "Infectious associations of celiac disease". *Autoimmunity Reviews* 8 (2009): 316-319. Volume 5 Issue 10 October 2022© All rights reserved by Parveen Malhotra., et al. Multiple Faces of Celiac Disease: A Case Report 24
8. Bentley D. A case of Down's syndrome complicated by retinoblastoma and coeliac disease. *Pediatrics* 1975; 56:131±133.
9. Simila S, Kokkonen J. Coexistence of coeliac disease and Down syndrome. *Am J Ment Retard* 1990; 95:120±122.
10. Granditsch G, Rossipal E. Down's syndrome and coeliac disease [Letter]. *J Pediatr Gastroenterol Nutr* 1990; 11:279.
11. Amil Dias J, Walker-Smith J. Down's syndrome and coeliac disease. *J Pediatr Gastroenterol Nutr* 1990; 10:41±43.
12. Santer R, Sievers E, Oldigs HD. coeliac disease in Down's syndrome [Letter]. *J Pediatr Gastroenterol Nutr* 1991; 13:121.
13. Hilhorst MI, Brink M, Wauters EA, Houwen RH. Down's syndrome and coeliac disease: A review of the literature. *Eur J Pediatr* 1993; 152:884±887.
14. Carnicer J, Crous A, Nosa S R, Artigas J, Lorente I. Dos casos de asociación de síndrome de Down y enfermedad celiac. *Pediatr Catalana* 1996; 56:43±45.
15. Rosso C, Cecere G, Concolino D, Baserga M. Diabetes, hypothyroidism and coeliac disease in Down's syndrome. A case report. *Minerva Pediatr* 1997; 49:483±485.
16. Storm W. Prevalence and diagnostic significance of gliadin antibodies in children with Down's syndrome. *Eur J Pediatr* 1990; 149:833±834.
17. Castro M, Crino A, Papadatou B, Purpura M, Giannotti A, Ferretti F, et al. Down's syndrome and coeliac disease: the prevalence of high IgA-antigliadin antibodies and HLA-DR and DQ antigens in trisomy 21. *J Pediatr Gastroenterol Nutr* 1993; 16:265±268.
18. Jansson U, Johansson C. Down syndrome and coeliac disease. *J Pediatr Gastroenterol Nutr* 1995; 21:443±445.
19. Failla P, Ruberto C, Pagano MC, Lombardo M, Bottaro G, Petichon B, et al. coeliac disease in Down's syndrome with HLA serological and molecular studies. *J Pediatr Gastroenterol Nutr* 1996; 23:303±306.
20. Zubillaga P, Vitoria JC, Arrieta A, Echaniz P, Garcia-Masdevall MD. Down's syndrome and coeliac disease. *J Pediatr Gastroenterol Nutr* 1993; 16:168±171.
21. Gale I, Wimalaratna H, Brotodiharjo A, Duggan JM. Down's syndrome is strongly associated with coeliac disease. *Gut* 1997; 40:492±496.
22. George EK, Mearin ML, Bouquet J, von Blomberg BM, Stapel SO, van Elburg RM, et al. High frequency of coeliac disease in Down syndrome. *J Pediatr* 1996; 128:555±557.

23. Carlsson A, Axelsson I, Borulf S, Bredberg A, Forslund M, Lindberg B, et al. Prevalence of IgA-antigliadin antibodies and IgA-ant endomysium antibodies related to coeliac disease in children with Down syndrome. *Pediatrics* 1998; 101:272±275.
24. Iacono G, Cavataio F, Balsamo V, Carroccio A, Montalto G, Notarbartolo A. coeliac disease in Down's syndrome: consideration on the role of histocompatibility antigens [Letter]. *J Pediatr Gastroenterol Nutr* 1990; 10:273.
25. Lundin KEA, Sollid LM, Qvigstad E. T lymphocyte recognition of a coeliac disease-associated cis- or trans-encoded HLA DQ alpha/beta heterodimer. *J Immunol* 1990; 145:136±139
26. McCulloch AJ, Ince PC, Kendall-Taylor P. Autoimmune chronic active hepatitis in Down's syndrome. *J Med Genet* 1982; 19:232±234.
27. Milunsky A, Neurath PW. Diabetes mellitus in Down's syndrome. *Arch Environ Health* 1968; 17:372±376.
28. Sare Z, Ruvalcaba RHA, Kelly VC. Prevalence of thyroid disorder in Down's syndrome. *Clin Genet* 1978; 14:154±158.
29. Catassi C, Fabiani E. The spectrum of coeliac disease in children. *Baillie; re Clin Gastroenterol* 1997; 11:485±507.
30. Calero P, Ribes-Koninckx C, Albiach V, Carles C, Ferrer J. IgA antigliadin antibodies as a screening method for non-overt coeliac disease in children with insulin-dependent diabetes mellitus. *J Pediatr Gastroenterol Nutr* 1996; 23:29±33.