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RESEARCH ARTICLE

CELIAC DISEASE AND DOWN SYNDROME

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Abstract

Celiac disease is immune related disorder of small bowel which is seen in genetically predisposed people and isdue to permanent intolerance to wheat gliadins and other cereal prolaminsalso known as gluten-sensitive enteropathy or non-tropical sprue. In 1888, it was first described byDr. Samuel Gee and in Greek means koiliakaos-abdominal. Dicke in1950 highlighted association between the consumption of bread, cereals and diarrhea which improved after stoppage of wheat intake. Thediagnosed 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.

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 prescence of glutamine and proline content in them in significant amount [2]. Inthe 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 prescence 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 rolein disease pathogenesis [6].The prevalence of CD worldwide and in India is globally 1%[7].Initially, association between Down's syndrome and coeliacdisease 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 withDown's syndrome, as reported in literature varies from 2.5 to 18.6% [16-23]

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Case Report

A fifteen-yearmale 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 withmild 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 IgATTG antibodies were massively raised to180 I.U./ml (normal being 0- 20 I.U./ml) and endoscopy showed severe scalloping of duodenalfolds 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 betweencoeliac disease and Down's syndrome, as reported in various studies [16-23]. Thus, whenever any new coeliac disease patient is diagnosed thenassociation with Down's syndrome should always be looked for. There is a highprevalence of immune-related disorders in patients withDown's syndrome, especially autoimmunethyroid disease, diabetes mellitus type I, autoimmune chronicactive hepatitis, alopecia, vitiligo, juvenile rheumatoidarthritisor 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 vigil 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.

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