

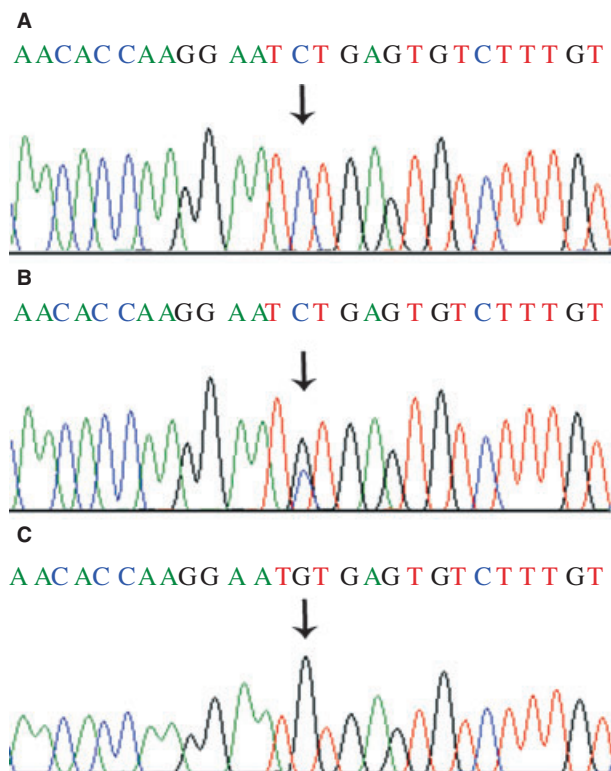


Figure 2. Sequence analysis of the *EDAR* gene mutation: Partial DNA sequence flanking the splice donor site of exon 5 of the *EDAR* gene from (A) a homozygous affected individual showing G to C transversion, (B) a heterozygous carrier, (C) a control individual showing normal splice donor site sequence GT. Arrows represent position of the mutation at splice donor site.

is necessary for normal development of ectodermal organs both in humans and mice. This study further supports the evidence of EDAR protein controlling the development/morphogenesis of ectodermal appendages.

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CHRONIC URTICARIA AND CELIAC DISEASE: A CASE REPORT

Abstract: We describe a case of a 9-year-old girl who presented chronic urticaria associated with celiac disease. The prevalence of the manifestation of chronic urticaria in celiac disease is unknown but increase in atopic immunologic disorders has been reported in the setting of gluten enteropathy. Relationship between the clinical manifestations is not clear. The present case of subclinical celiac disease diagnosis in an otherwise asymptomatic child with chronic urticaria further reinforces the evidence that differential for celiac disease warrants to be always considered in children with refractory urticaria.

A 9-year-old female patient was admitted to our Clinics for chronic urticaria (CU). Hematologic and routine biochemistry blood tests were within normal ranges as well as microbiological investigations. Chest radiograph and abdominal echo scan were unremarkable. All skin prick tests to foods and inhalants resulted negative. Anti-transglutaminase (21.00 UA/L) and anti-endomysial

antibodies were found positive. Endoscopic test with bowel biopsy demonstrated villous atrophy, dense plasma cells and increase of intraepithelial lymphocytes. Diagnosis of celiac disease (CD) associated with CU was performed. Patient has begun a gluten-free diet, which was followed by prompt resolution of urticaria manifestations.

Chronic urticaria may be related to inflammatory gastrointestinal diseases or autoimmune disorders (autoimmune thyroiditis, type I diabetes mellitus, alopecia) that often are under-diagnosed. Celiac disease, or gluten sensitive enteropathy, is an inflammatory condition of the small intestine, induced by gluten, which is derived from wheat, barley, and rye. In patients with CD, immune responses to gliadin fractions promote an inflammatory reaction, primarily in the upper small intestine, characterized by infiltration of the lamina propria and the epithelium with chronic inflammatory cells and villous atrophy. The symptoms vary ranging from diarrhea, abdominal distension, and failure to thrive in infants, vomiting, irritability, anorexia, and even constipation in older children, to extra-intestinal manifestations, such as short stature, neurologic symptoms, or anemia in adolescents. The relationship (1,2) between the two diseases is not clear but it can be speculated that autoimmunity induced by gliadin or by other unknown antigens may link CU and CD. The increased permeability of intestinal mucosa (3) allows the passage of antigens that are responsible for CU pathogenesis by the formation of circulating immuno-complexes. Gliadin peptide antigen processing causes the production of IgA and IgG which can lead to the formation of C5a, an anaphylotoxin able to induce mast cells and basophil degranulation and release of mediators. It is tempting to speculate that this mechanism could be responsible for CU manifestation, and that skin symptoms could resolve by restoring the integrity of the mucosa after a gluten-free diet. Nevertheless, both CD and urticaria are immune-mediated disorders, but they have a different pathogenesis; CD is a TH₁-mediated autoimmune response to gluten while urticaria could be supported by different mechanisms that range from TH₂-driven response to allergens to TH₁-autoimmunity (1). CU is related to autoantibodies against the α -subunit of the high affinity IgE receptor Fc ϵ R1 or against the α subunit of IgE. These antibodies are able to induce the mast cells degranulation and the consequent formation of anaphylatoxin (4).

It has been reported that patients with CU have higher prevalence of CD (1). In most of these reports, an improvement in CU after starting gluten-free diet is described (1) and it has hypothesized that a reduction in the immunologic stimulus may account for symptom

improvement in children with subclinical CD associated with CU (5). The present case of subclinical CD diagnosis in an otherwise asymptomatic child with CU further reinforces evidence that differential for CD warrants to be always considered in children with refractory CU.

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CONGENITAL LUPUS ERYTHEMATOSUS PRESENTING AT BIRTH WITH WIDESPREAD EROSIONS, PANCYTOPENIA, AND SUBSEQUENT HEPATOBILIARY DISEASE

Abstract: Neonatal lupus erythematosus is an uncommon disease caused by transplacental passage of maternal anti-Ro (SS-A), anti-LA (SS-B), or anti-U1RNP antibodies. Cutaneous findings of neonatal lupus are variable, but annular, erythematous plaques occurring within a few weeks of birth are most typical. Cutaneous lesions of congenital onset lupus erythematosus can differ from that of neonatal lupus erythematosus, presenting with atrophy or scarring, and less commonly, erosions. We report an unusual case of congenital lupus erythematosus presenting at birth with widespread erosions, pancytopenia, and subsequent hepatobiliary disease.
