

# Clinical Scenario for Medical Students in Pediatric Multiple Autoimmune Diseases

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**Abstract** In the accordance of readers, we are introducing a new chapter in Journal, which will consist in Clinical Scenarios for Medical Students. First clinical scenario is presented by associate professor Sur Genel and his team: a 12-year-old girl diagnosed with a combination of four autoimmune diseases: autoimmune hepatitis, thyroiditis, celiac disease and type 1 diabetes mellitus.

**Keywords:** multiple autoimmune syndrome, polyautoimmunity

**Cite This Article:** Sur M Lucia, Floca Emanuela, Sur Daniel G, Lupan Iulia, Sur Genel, Samasca Gabriel, and Aldea Cornel, "Clinical Scenario for Medical Students in Pediatric Multiple Autoimmune Diseases." *International Journal of Celiac Disease*, vol. 5, no. 4 (2017): 171-172. doi: 10.12691/ijcd-5-4-9.

## 1. Introduction

Autoimmune disease (AD) is a pathologic condition caused by loss of immunological tolerance to self-antigens [1]. The balance between self-tolerance and autoimmune genes is involve many self-protective immune mechanisms that try to suppress pro-inflammatory processes [2]. Multiple ADs are associated with each other and may converge to one individual or pile up in the same family that acquired, the so called the "autoimmune trait". Parents with a personal history of an AD have a higher risk to pass on the autoimmunity trait to their offspring. Polyautoimmunity is defined as the presence of more than one AD in a single patient. When three or more ADs coexist, this condition is called multiple autoimmune syndrome, which represents an interesting and challenging model to explore the cross-talks between genes and environment in the autoimmune puzzle [3]. Studies have shown that about 25 percent of patients with ADs can develop additional autoimmune diseases [4,5].

## 2. Case Report

A 12-year-old girl came to our medical pediatric facility, in Romania, for work-up. From the past medical history, her mother recalled that her daughter presented, around the age of one, a capricious appetite, bulky pasty stools and poor weight gain. Her pediatrician didn't evaluate her, but instead, gave her symptomatic therapy and suggested to follow her up. In the same time, the baby girl developed a recurrent skin rash. From 1 to 6 years of age she had neither hospitalization nor any investigation.

At the age of 6 years she was admitted in the pediatric service for the investigation of a hepatic cytolysis syndrome, manifested by transaminasemia. At the same time, she developed peripheral facial paralysis, accompanied by a surge in hepatic transaminases. It is worth mentioning that the celiac disease associated antibodies were fluctuating, between positive and negative titers, IgA-transglutaminase and IgA-anti endomysial were never positive at the same time.

Laboratory investigations conducted at the age of 6 years were normal for: HBV, HAV, HCV, HDV, CMV, and EBV. Autoimmune hepatitis was initially ruled out by the lack of inflammatory markers, Ac-ANA, p-ANCA and Transglutaminase antibodies were equally negative.

Between 6 -10 years of age, the girl was investigated by her community pediatrician, in Italy, but no progress was done concerning the etiology. At 10 years old, she came back to the pediatric service of Cluj-Napoca, Romania, for investigations. IgA-Transglutaminase antibodies, at that time, were weakly positive. Abdominal ultrasonography suspected liver cirrhosis. Despite it, the parents refused liver and duodenal biopsy. Fortunately, in the same year, investigations conducted in Italy disclosed autoimmune hepatitis in cirrhotic stage with fibrosis degree IV. Duodenal biopsy was compatible with the diagnosis of celiac disease with stage 3C Marsh, degree of atrophy. Patient started gluten-free diet for celiac disease and cortisone and Imuran, for her advanced autoimmune hepatitis. Hepatic functions, weight and stools and abdominal pains, ameliorated.

At the age of 12 the patient returned for investigation, at which point she was euthyroidic, but positive for anti-thyroid autoantibodies. She was diagnosed with the third AD: celiac disease, autoimmune hepatitis and presently, autoimmune thyroiditis. On follow-up, her fasting serum

glucose levels oscillated between 100-130mg / dL. She had mild glycosuria. An abnormal glucose tolerance test was detected and her Glycosylated hemoglobin (HbA1c) was 6%. The fourth AD was added to the three and anti-diabetes mellitus type 1 therapy was initiated.

### **3. Conclusion**

What is the main take home message from the present case report?

### **References**

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