Abstract 991

AUTOIMMUNE HEPATITIS AND IMMUNE DYSREGULATION: A CASE SERIES

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Authors: Giulia Jannone, Clément Triaille, Fabien Touzot, Fernando Alvarez; CHU Sainte-Justine, Montreal, Canada

Objectives and Study

To assess the presence of inborn errors of immunity (IEI) in a pediatric autoimmune hepatitis (AIH) cohort.

Methods

This retrospective study included all patients aged 0-18 diagnosed with AIH in our center between 1995 and 2023 and concurrently followed by the immunology department for a clinical and/or molecular IEI diagnosis.

Results

Amongst the 83 patients followed in our center for AIH, six patients (7.2%) displayed signs of associated IEI. Two of those six patients had a genetic confirmation of IEI (*SP110* and *AIRE* homozygous mutations), three patients displayed variants of uncertain significance in immunodeficiency-related genes, and the last patient was not genetically tested. IEI-related signs were recurrent infections (n=4), immune-mediated cytopenia (n=4), immune-mediated skin disease (n=3), chronic diarrhea (n=1) and autoimmune polyendocrinopathy (n=1). Four patients were diagnosed with seronegative AIH and two patients with AIH type 2. All patients responded to AIH immunosuppressive therapy, except for the two AIH type 2 patients who underwent emergent liver transplantation for fulminant liver failure at diagnosis.

Conclusions

Our small case series highlights the need to look for signs of immune dysregulation in AIH patients. Conversely, as AIH can be atypical in IEI, the threshold should be low to perform a diagnostic liver biopsy in IEI patients suffering from chronic cytolysis. We believe that systematic genetic testing and immune phenotyping of AIH patients who display signs of dysimmunity will be crucial to better understand the close link between AIH and IEI.

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