

A 12-year-old Boy with Generalized Edema

A 12-year-old boy who was referred with generalized edema, nephrotic range proteinuria, hypoalbuminemia and hyperlipidemia. His laboratory investigations showed thrombocytopenia and mild anemia, the other lab tests like kidney function test, liver function tests, complements, antinuclear antibody and serum electrolytes were in normal range. His urinalysis showed normal data except proteinuria. His physical examination revealed patchy skin lesions on dorsal side of the hands and feet (figure 1). Clinical signs and symptoms of the patient improved after 6 weeks of corticosteroids usage.

What is your diagnosis?



A 12-year-old Boy with Generalized Edema

Mojtaba Fazel*, Negin Sadat Valiahdi

Department of Pediatrics, Valiasr Hospital, Imam Khomeini Medical Complex, Tehran University of Medical Science, Tehran, Iran

Please cite this article as: Fazel M, Valiahdi N. A 12-year-old Boy with Generalized Edema. J Ped Nephrol 2020;8(1):1-2.
<https://doi.org/10.22037/jpn.v8i1.30006>

***Corresponding Author**

Mojtaba Fazel, MD.

Email: mojtabafazel@yahoo.com

Final diagnosis of the patient based on genetic study was Wiskott-Aldrich syndrome (WAS). WAS is an X-linked disorder caused by mutations in the gene that encodes the Wiskott-Aldrich syndrome protein (WASp). The originally described features of WAS include susceptibility to infections (subsequently associated with adaptive and innate immune deficiency), microthrombocytopenia, and eczema.

The pathogenesis of renal involvement in Wiskott-Aldrich syndrome (WAS) is unclear and renal outcome is generally poor in such situations.

References:

1. Ochs HD, Thrasher AJ. The Wiskott-Aldrich syndrome. Journal of Allergy and Clinical Immunology 2006;117(4): 725-738.
2. F Candotti. Clinical Manifestations and Pathophysiological Mechanisms of the Wiskott-Aldrich Syndrome. Journal of clinical immunology 2018.
3. Tomonari Shigemura, Yozo Nakazawa, Hisashi Shimojo, et al. Immune Complex-Mediated Glomerulonephritis in a Patient with Wiskott-Aldrich Syndrome. Journal of Clinical Immunology 2016;36:357-359.