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Excellent Outcome with Early Diagnosis in an Infant with Wiskott-Aldrich Syndrome in a Tertiary Hospital in Oman

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Abstract : Wiskott-Aldrich syndrome (WAS) is a primary immunodeficiency disease resulting in recurrent infections, eczema, and microthrombocytopenia. In its classical form, significant combined immune deficiency, autoimmune complications, and risk of hematological malignancy necessitate early correction, preferably before 2 years of age, with hematopoietic stem cell transplant (HSCT) or gene therapy. Clinical features and severity are varied, making the diagnosis difficult in milder cases. We report an Omani boy diagnosed in early infancy with WAS based on clinical presentation and confirmed by genetic diagnosis with cure by HSCT from an HLA-identical sibling donor.

Keywords : genetic diagnosis, hematopoietic stem cell transplant, infant, Wiskott-Aldrich syndrome
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