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Persistent Microthrombocytopenia with Eczema: Dermatologic Diagnosis of Wiskott–Aldrich Syndrome

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Abstract:

Background:
Wiskott–Aldrich syndrome (WAS) is a rare X-linked primary immunodeficiency characterized by the triad of microthrombocytopenia, eczema, and recurrent infections. Early manifestations are frequently misdiagnosed as immune thrombocytopenic purpura (ITP), resulting in delayed definitive diagnosis and management. Recognition of persistently reduced mean platelet volume (MPV), particularly in association with chronic eczema, is critical for early and accurate diagnosis.
Case Presentation:
We report a 6-year-old boy who initially presented at 9 months of age with recurrent febrile episodes and spontaneous bleeding manifestations, including epistaxis, hematemesis, hematochezia, petechiae, and ecchymoses. Laboratory evaluation revealed persistent thrombocytopenia with markedly reduced MPV (<5 fL). Bone marrow examination demonstrated normocellular marrow with increased megakaryocytes, leading to an initial diagnosis of ITP and treatment with corticosteroids and intravenous immunoglobulin, resulting in only transient improvement. Over time, he developed progressive, treatment-refractory eczematous dermatitis with hemorrhagic features, recurrent infections, and failure to thrive. Serial hematologic assessments consistently demonstrated microthrombocytopenia, prompting evaluation for an inherited thrombocytopenic disorder. Targeted next-generation sequencing identified a hemizygous pathogenic c.257G>A (p.Arg86His) variant in the WAS gene, confirming classical WAS. He received supportive dermatologic and hematologic management and was referred for hematopoietic stem cell transplantation (HSCT); however, transplantation was deferred due to financial constraints.
Conclusion:
Persistent microthrombocytopenia with reduced MPV in male infants, particularly when accompanied by chronic eczema and recurrent infections, should prompt evaluation for Wiskott–Aldrich syndrome. Early recognition through careful interpretation of platelet indices and dermatologic assessment is essential to avoid misdiagnosis as ITP and to facilitate timely molecular confirmation and referral for definitive therapy. Heightened multidisciplinary awareness is essential to reduce diagnostic delay and optimize long-term outcomes.

Keywords:

Wiskott–Aldrich syndrome; microthrombocytopenia; mean platelet volume; eczema; inherited thrombocytopenia; primary immunodeficiency

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Introduction

Wiskott–Aldrich syndrome (WAS) is a rare, inherited, X-linked primary immunodeficiency disorder characterized by the classical triad of microthrombocytopenia, eczema, and recurrent infections [1]. First described in 1937 by Alfred Wiskott and later recognized as an X-linked condition by Robert Aldrich, WAS is now established as a distinct genetic syndrome affecting immune cell function [1, 2]. The estimated incidence is approximately 1–4 per million live male Thrasher births worldwide [3,4].

WAS results from mutations in the *WAS* gene located on chromosome Xp11.23, which encodes the Wiskott–Aldrich syndrome protein (WASP), a critical regulator of actin cytoskeleton remodeling in hematopoietic cells [5]. Dysfunction of WASP disrupts T-cell, B-cell, natural killer cell, and dendritic cell activity, leading to combined immunodeficiency. Clinically, patients typically present in early infancy with persistent thrombocytopenia and characteristically small platelets, resulting in bleeding manifestations such as petechiae, purpura, and mucosal hemorrhage, along with eczematous dermatitis and recurrent infections [1, 3]. Autoimmune complications and malignancies may also develop. The phenotypic spectrum ranges from classic WAS to milder variants such as X-linked thrombocytopenia. Diagnosis is established by demonstrating reduced or absent WASP expression and confirmatory molecular genetic testing [1,5, 6].

Although supportive management remains essential, hematopoietic stem cell transplantation is currently the only established curative therapy, with optimal outcomes when performed early [1]. Given the potential for diagnostic delay due to overlapping dermatologic and hematologic features, early recognition is critical. We report the case of a 6-year-old boy presenting with persistent microthrombocytopenia and eczema, underscoring the importance of dermatologic manifestations in facilitating timely diagnosis of WAS.

Case Presentation

A 6-year-old boy, born to non-consanguineous parents, initially presented at 9 months of age with recurrent febrile episodes and spontaneous bleeding manifestations, including epistaxis, hematemesis, hematochezia, and multiple petechiae and ecchymoses over the trunk and extremities. Initial laboratory investigations revealed thrombocytopenia with persistently reduced mean platelet volume (MPV). Bone marrow examination demonstrated normocellular marrow with increased megakaryocytes, without evidence of marrow failure. A provisional diagnosis of immune thrombocytopenic purpura (ITP) was made, and the child received multiple courses of systemic corticosteroids, dapsone, intravenous immunoglobulin (IVIG), and platelet transfusions. These therapies resulted only in transient improvement, followed by rapid relapse of thrombocytopenia and recurrent bleeding episodes.

Over the subsequent 1–2 years, the patient developed progressively worsening eczematous dermatitis. Dermatologic examination revealed multiple erythematous, edematous plaques with excoriations, crusting, oozing, and scaling involving the bilateral lower limbs, knees, flexural aspects of the upper limbs, and inguinal folds. Areas of lichenification and post-inflammatory hyperpigmentation indicated chronicity. Numerous petechiae and purpuric macules were present within and surrounding the eczematous lesions, as well as on clinically uninvolved skin, reflecting the underlying bleeding diathesis. Representative clinical photographs demonstrating the evolution of the cutaneous findings at ages 3, 6, and 7 years are shown in **Figure 1A–C**.

The child experienced recurrent hospitalizations for upper respiratory tract infections, acute gastroenteritis, intermittent febrile illnesses, and management of bleeding complications. Growth assessment demonstrated failure to thrive with stunting persisting up to 6 years of age. There was no history of early male sibling deaths, similar dermatologic or hematologic conditions, or known primary immunodeficiency in the family.

Serial hematologic evaluations consistently demonstrated thrombocytopenia, with platelet counts ranging from 9,000 to 52,000/µL. MPV remained persistently low (<5 fL), indicative of microthrombocytopenia. Peripheral blood smear confirmed markedly reduced platelet numbers with

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predominantly small-sized platelets and associated microcytic hypochromic anemia. The persistently reduced MPV was atypical for ITP, in which platelet size is typically normal or increased, raising suspicion for an inherited thrombocytopenic disorder. Routine biochemical investigations were within reference limits.

Given the triad of early-onset microthrombocytopenia, chronic recalcitrant eczema with hemorrhagic features, and recurrent infections, Wiskott–Aldrich syndrome was strongly suspected. The patient was referred for immunologic and molecular evaluation. Targeted next-generation sequencing identified a hemizygous c.257G>A (p.Arg86His) missense variant in the *WAS* gene, previously reported as pathogenic and consistent with the classical WAS phenotype. A WAS clinical score of 3, together with molecular confirmation, established the diagnosis.

Cutaneous manifestations were managed with gentle skin care measures, regular emollient therapy, and intermittent topical corticosteroids, with topical antibiotics during exudative flares. Systemic antimicrobial therapy was administered for intercurrent infections, and platelet transfusions were provided during significant bleeding episodes. The family received genetic counseling regarding the X-linked inheritance pattern, carrier screening for at-risk female relatives, and the role of hematopoietic stem cell transplantation (HSCT) as definitive therapy. Although referred for HSCT evaluation, transplantation was deferred due to financial constraints. At the most recent follow-up, the child remained under multidisciplinary care, with improved control of eczema but persistent thrombocytopenia and ongoing risk of bleeding complications.

Figure 1. Progressive eczematous dermatitis in Wiskott–Aldrich syndrome.

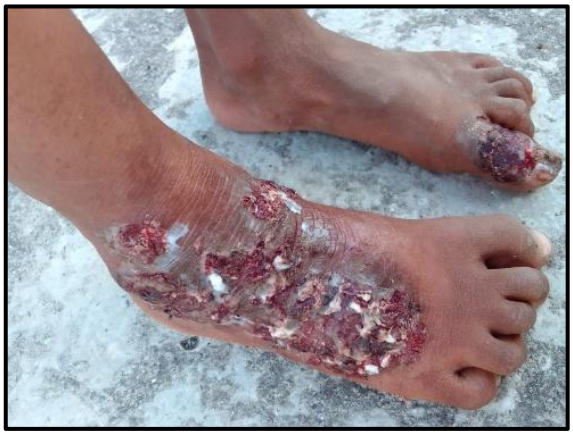

Figure 1. (A) Age 3 years: Erythematous to hyperpigmented plaques with erosions, crusting, and pustulation over the right dorsum of the foot and left great toe.

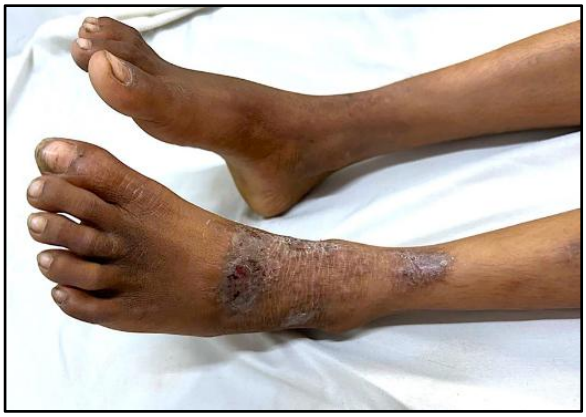

Figure 1. (B) Age 6 years: Hyperpigmented to erythematous plaques with superficial erosions and scaling involving the left dorsum of the foot and lower limb.

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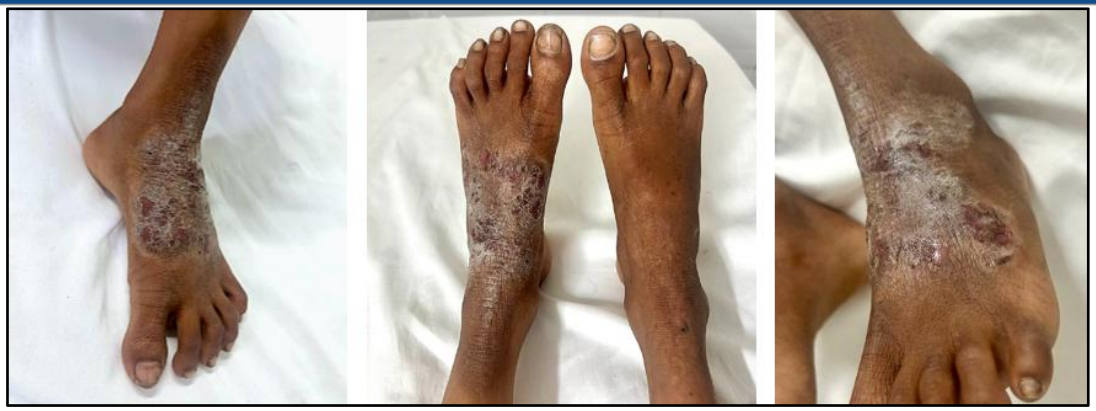

Figure 1. (C) Age 7 years: Chronic hyperkeratotic plaques with persistent erosions and scaling over the left dorsum of the foot, reflecting disease chronicity.

Discussion

Wiskott–Aldrich syndrome (WAS) remains an under-recognized cause of syndromic thrombocytopenia in male infants, particularly when early-onset thrombocytopenia is presumed to be immune-mediated. Mutations in the *WAS* gene impair cytoskeletal signaling in hematopoietic cells, leading to microthrombocytopenia and progressive immune dysregulation. As emphasized by Sullivan KE et al (1994) [7] and Suri D et al (2021) [4], bleeding manifestations and eczema are present in the majority of patients with classical WAS and frequently precede overt immunologic deterioration. Buchbinder D et al (2014) [8] further underscored that early diagnosis is critical, as definitive therapy is time-sensitive and outcomes are strongly influenced by the timing of intervention.

A major diagnostic challenge, illustrated in this case, is misclassification as immune thrombocytopenic purpura (ITP). In infancy, isolated thrombocytopenia commonly prompts empiric treatment for ITP, particularly when bone marrow examination demonstrates preserved cellularity with increased megakaryocytes. However, persistently reduced mean platelet volume (MPV) represents a key distinguishing feature of WAS. Unlike ITP—where platelet size is typically normal or increased due to compensatory megakaryopoiesis—WAS is characterized by microthrombocytopenia with consistently small platelets. Similar diagnostic delays have been reported by Dutta A et al (2025) [1] and Gaikwad P et al (2024) [9], reinforcing the importance of meticulous evaluation of platelet indices and peripheral smear morphology. Early-onset thrombocytopenia with persistently low MPV should therefore prompt evaluation for inherited thrombocytopenic disorders.

Beyond hematologic abnormalities, dermatologic manifestations may provide a critical diagnostic clue. WAS-associated eczema frequently mimics atopic dermatitis but is typically persistent and often refractory to conventional therapy. Cohort analyses by Suri D et al (2021) [4] report cutaneous involvement in approximately 80% of classical cases. In the present patient, the coexistence of petechiae and purpura within eczematous plaques suggested an underlying bleeding diathesis, distinguishing this presentation from isolated inflammatory dermatoses. Recognition of the combined phenotype of chronic eczema and microthrombocytopenia is essential to facilitate early referral and comprehensive immunologic assessment.

Regional cohort data from India further demonstrate heterogeneous clinical presentations, frequent diagnostic delay, and limited access to definitive therapy in resource-constrained settings, as summarized by Pilania RK et al (2020) [10] and Suri D et al (2021) [4]. Mutational analyses reported by Gaikwad P et al (2024) [9] highlight significant genotypic heterogeneity, underscoring the importance of molecular confirmation for accurate diagnosis and prognostic stratification. Delayed recognition carries substantial risk, including autoimmunity and lymphoid malignancy, as described by Senapati J et al (2014) [11], emphasizing the prognostic implications of early identification.

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Hematopoietic stem cell transplantation (HSCT) remains the established curative therapy for classical WAS. Registry data from Ferrua F et al (2019) [12] and Burroughs LM et al (2020) [13] demonstrate significantly improved survival when transplantation is performed early in the disease course. Emerging gene therapy approaches, initially reported by Braun CJ and subsequently refined by Moratto D et al (2011) [14], have shown promising long-term immune reconstitution and represent an evolving therapeutic alternative for selected patients.

This case reinforces several important clinical principles: persistent thrombocytopenia with reduced MPV in male infants warrants evaluation for inherited thrombocytopenia; chronic eczema accompanied by hemorrhagic features should raise suspicion of underlying immune dysregulation; and early molecular confirmation is essential to enable timely referral for definitive therapy. Heightened multidisciplinary awareness is imperative to reduce diagnostic latency and optimize survival, immune reconstitution, and long-term outcomes in children with Wiskott–Aldrich syndrome.

Conclusion

This report underscores the diagnostic significance of persistent microthrombocytopenia in conjunction with chronic eczema as an early clinical indicator of Wiskott–Aldrich syndrome. Failure to recognize the characteristic finding of reduced mean platelet volume may lead to misclassification as immune thrombocytopenic purpura and consequent delay in definitive diagnosis. The case highlights the pivotal role of meticulous dermatologic evaluation in raising suspicion for an underlying primary immunodeficiency, particularly when cutaneous manifestations are accompanied by recurrent infections and bleeding diathesis.

Early identification, supported by characteristic hematologic parameters and confirmed through molecular testing, is critical to ensure timely referral for definitive management, including hematopoietic stem cell transplantation. Greater awareness among pediatricians, dermatologists, and hematologists is essential to promote prompt diagnosis, appropriate genetic counseling, and improved long-term clinical outcomes.

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