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LESSONS

Neonatal Thrombocytopenia and Rectal Bleeding as Initial Manifestations of Wiskott-Aldrich Syndrome

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A male infant was born via lower segment cesarean section (LSCS), to a G2P1L1 mother with a birth weight of 3.78 kg. On day 3 of life, he developed periumbilical redness with a platelet count of 20,000/µL, prompting platelet transfusions. Workup for thrombocytopenia was done. On day 8, he had blood in stools with platelets at 22,000/µL. He received another transfusion and intravenous immunoglobulin (IVIG) and methylprednisolone (for 5 days) were administered. CMV PCR and genotyping for neonatal alloimmune thrombocytopenia (NAIT) were negative. Whole exome sequencing (WES) suggested Wiskott-Aldrich Syndrome (WAS).

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The baby was readmitted at 3 months of age in view of bleeding and received IVIG and platelet transfusions, and was discharged. After 2 weeks, he presented to the emergency department with bleeding per rectum approximately 12 hours prior to the current admission, with a platelet count of 9,000/µL. On examination, baby had signs of eczema with mild erythematous maculopapular rash, scaling on the forehead and scalp, along with scattered petechiae (Figure 1). Two units of platelet transfusions were given, after which bleeding stopped. Repeat complete blood count (CBC) showed platelets at 115,000/µL (1.15 lacs/µL). No fresh bleeding was noted post-admission. Baby is now planned for a hematopoietic stem cell transplant (HSCT).

This case highlights early neonatal presentation of WAS with isolated thrombocytopenia and bleeding, confirmed by WES after exclusion of common differentials like infection and NAIT. In

acute episodes, stabilization can be achieved by supportive care including transfusions and IVIG. The infant is receiving under multidisciplinary follow-up for potential immunodeficiency, eczema, autoimmunity, and simultaneously is being planned for definitive therapy (hematopoietic stem cell transplantation-HSCT).

Figure 1. Clinical photographs of the male infant with Wiskott-Aldrich syndrome at approximately 3 months of age. (A) Close-up facial view showing signs of eczema with mild erythematous maculopapular rash, scaling on the forehead and scalp, along with scattered petechiae. (B) Full-body view demonstrating generalized skin involvement with eczematous changes and scaling, crying, and petechial/purpuric lesions consistent with thrombocytopenia. The infant presented with neonatal thrombocytopenia and rectal bleeding; diagnosis was confirmed by whole exome sequencing. Images taken during hospital admission for an acute rectal bleeding episode.

Lessons

Wiskott-Aldrich Syndrome is a rare X-linked primary immunodeficiency caused by mutations in the *WAS* gene. This gene is located on chromosome Xp11.22-23 and encodes the WAS protein (WASp) [1,2]. WASp protein regulates actin cytoskeleton reorganization in hematopoietic cells and is instrumental in immune synapse formation, cell migration, phagocytosis, and platelet function. Mutations in this gene can lead to a spectrum of phenotypes: classic, milder X-linked thrombocytopenia (XLT), and X-linked neutropenia (XLN). Absent WASp expression typically causes classic disease, while residual expression correlates with milder forms [2,3] (Table 1).

Table 1. Phenotypic Spectrum of WAS-Related Disorders

	Pathophysiology	Clinical features
Classic WAS	Absent WASp	Microthrombocytopenia, eczema, infections, autoimmunity, malignancy risk.
XLT	Residual WASp	Milder thrombocytopenia, fewer infections.
XLN	Gain-of-function mutations	Neutropenia, variable other features.

Clinical Presentation and Diagnosis

Thrombocytopenia with small platelets is often the earliest and most consistent feature, present from birth in many cases. Neonatal bleeding (e.g., petechiae, umbilical oozing, bloody stools, or post-circumcision hemorrhage) is common, as seen here. Eczema and infections may appear later or be absent initially. Figure 2 shows the classic clinical features of WAS in an infant. The differential diagnoses of WAS in neonates are NAIT, congenital infections (e.g., CMV), sepsis, and other inherited thrombocytopenias [4,5]. Diagnosis was made in this case by negative NAIT genotyping and CMV PCR, followed by WES. Flow cytometry for WASp expression in lymphocytes can serve as a rapid screen for diagnosis before genetic confirmation. Platelet size (mean platelet volume) is also reduced, although some variants show normal size.

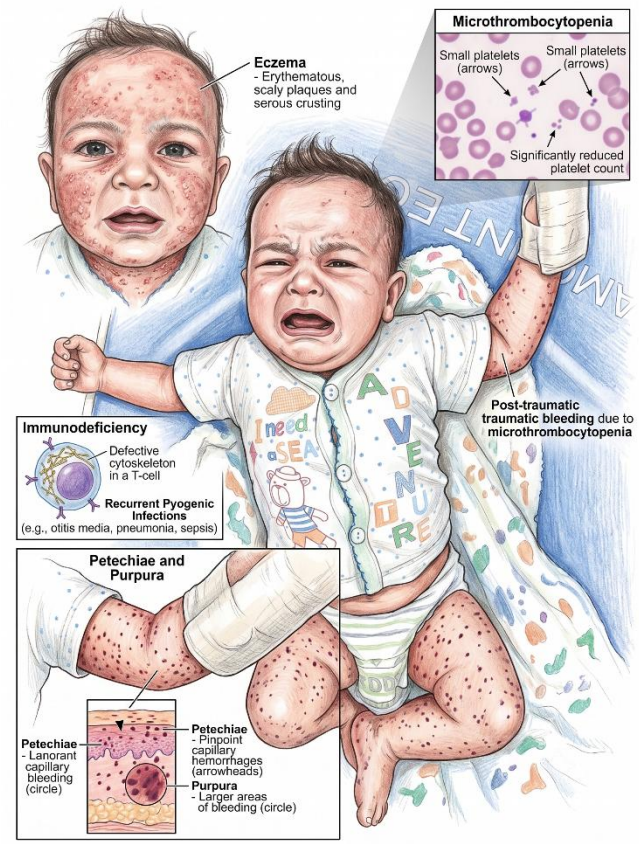

Figure 2. Schematic illustration depicting the classic clinical and laboratory features of Wiskott-Aldrich syndrome (WAS) in a neonate/infant.

Early diagnosis via WES is indispensable in ambiguous neonatal thrombocytopenia, which reduces delays in diagnosis and enables timely intervention. This case underscores the utility of genomic sequencing in refractory or atypical thrombocytopenia, or male predominance.

Management Challenges

Acute bleeding episodes mandate platelet transfusions, though responses can be transient due to intrinsic platelet dysfunction and immune clearance. IVIG does benefit in some cases by modulating autoimmunity and supporting humoral immunity. Corticosteroids were used in our case for presumed immune-mediated components [6]. Prophylactic antibiotics, IVIG transfusion, and eczema management are supportive. Splenectomy may theoretically raise platelet counts but leads to elevated risk of infections and is generally avoided or combined with lifelong prophylaxis.

The definitive treatment is allogeneic HSCT, ideally from a matched sibling or unrelated donor, with excellent outcomes when performed early (before complications like autoimmunity or malignancy) [7]. Gene therapy is an emerging option for patients lacking suitable donors [8]. Patients should be followed up for autoimmunity (e.g., hemolytic anemia, vasculitis, IBD) and malignancy (especially EBV-associated lymphoma) because these conditions can contribute significantly to morbidity.

Prognosis and Broader Implications

Survival is limited in the absence of HSCT due to bleeding, infections, or malignancy [9]. Outcomes are improved by early diagnosis and treatment. This case shows that WAS can present with predominant hematologic features in the neonatal period and can mimick other hematologic ailments. Neonatologists should maintain a high index of suspicion for primary immunodeficiencies in male neonates with persistent thrombocytopenia and bleeding which responds poorly to standard measures. Multidisciplinary care involving hematology, immunology, and genetics is indispensable in managing a case of WAS.

Statements and Declarations

Conflicts of interest

The authors declare that they do not have conflict of interest.

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