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Wiskott-Aldrich Syndrome: Case Report

Mary Minolin. T[1], Praveen Raj M[2]

[1]Professor, Department Child Health Nursing, Saveetha College of Nursing,
[2]MSc (Nursing), NPCC (Nurse Practitioner in Critical Care), Saveetha College of Nursing,
[1,2]Saveetha Institute of Medical and Technical Sciences, Saveetha University, Chennai, Tamil Nadu, India

ABSTRACT

Wiskott-Aldrich syndrome is characterized by abnormal immune system function (immune deficiency), eczema (an inflammatory skin disorder characterized by abnormal patches of red, irritated skin), and a reduced ability to form blood clots. This condition primarily affects males. Individuals with Wiskott-Aldrich syndrome have micro thrombocytopenia, which is a decrease in the number and size of blood cells involved in clotting (platelets). This platelet abnormality, which is typically present from birth, can lead to easy bruising, bloody diarrhoea, or episodes prolonged bleeding following nose bleeds or minor trauma. A 9-month-old boy baby presented with the report of a small red rash consisting of "dots" under the skin (these are called petechiae, frequent and easy bleeding that can occur in bowel movements.

KEYWORDS: *wiskott Aldrich syndrome, petechiae, bloody diarrhea*

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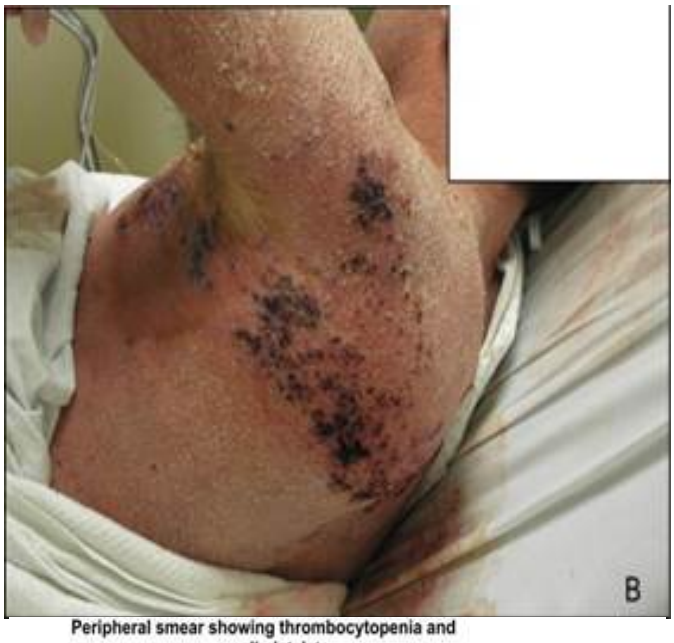
INTRODUCTION

It is a disease with immunological deficiency and reduced ability to form blood clots. Signs and symptoms include easy bruising or bleeding due to a decrease in the number and size of platelets; susceptibility to infections and to immune and inflammatory disorders; and an increased risk for some cancers (such as lymphoma). Also, a skin condition known as eczema is common in people with WAS. Wiskott Aldrich syndrome is caused by genetic changes in the WAS gene and is inherited in an X-linked manner. It primarily affects males. Wiskott-Aldrich syndrome, X-linked thrombocytopenia (XLT), and X-linked neutropenia (XLN) are known as 'WAS-related disorders' because these diseases are all caused by genetic changes in the WAS gene, and have overlapping symptoms ranging from severe to mild (Wiskott-Aldrich syndrome is the most severe)

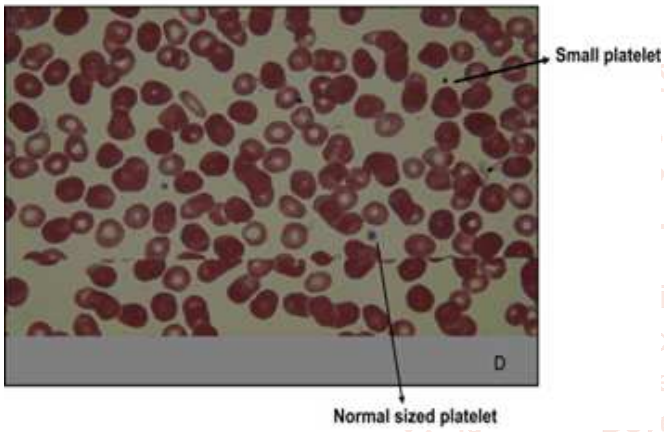
Case Description:

A 9-month-old boy baby was referred for further investigations in view of persistent thrombocytopenia from birth. He was admitted to the neonatal ward after birth due to borderline prematurity at 35 weeks. A full blood count taken on the first day of life (DOL 0) showed thrombocytopenia with a platelet count of 43,000/µL and a normal mean platelet volume (MPV) of 8.4 fL. Results from a TORCH screen (for *Toxoplasma gondii*, other viruses, rubella, cytomegalovirus, and herpes simplex) were negative. He was treated for presumed sepsis. His platelet count while on our ward ranged from 25,000/µL to 68,000/µL.

After discharge, his platelet counts during follow-up appointments at the clinic ranged from 20,000/µL to 30,000/µL and no bleeding tendency was reported. Our patient was the only male among his siblings. His parents were non-consanguineous. Two of his maternal uncles died during infancy from unknown causes (at ages 1 month and 7 months). None of them was investigated for WAS. A mutation analysis of our patient revealed a c.1264G>T mutation in exon 10 of the *WAS* gene, which led to a change in the 422nd amino acid from alanine to serine.



Peripheral smear showing thrombocytopenia and small platelets



Discussion:

Wiskott–Aldrich Syndrome (WAS) is a rare X-linked recessive disease that was first recognized as a clinical syndrome comprising of immunodeficiency, thrombocytopenia, bloody diarrhea and eczema by Wiskott in 1937. The X-linked mode of inheritance was subsequently demonstrated by Aldrich. The incidence of WAS is estimated between 1 and 10 in 1 million live births, although this is likely to be an underestimation, as patients lacking the classic phenotype are often unrecognized.

The number of B cells might be normal or moderately decreased. Serum IgM levels are moderately decreased or can be normal and increased. IgA and IgE levels are frequently increased whereas serum IgG levels are within normal range. These immune deficiencies lead to recurrent bacterial and viral infections.

Conclusions:

This Case demonstrates the importance of complete medical and family history as well as necessity of a multidisciplinary approach and a broad differential diagnosis when a congenital disease in adults is suspected. Although there are few reports of WAS in adults, one should consider this diagnosis in an appropriate clinical setting because of variable penetrance of the disease.

Consent for publication: Informed consent was obtained from parents of the patients to publish this case in medical journal.

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Conflict of interest: None declared

Ethical approval: Not required

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