

Wiskott-Aldrich Syndrome: Case Report

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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 diarrhoea*

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/ μ L 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/ μ L to 68,000/ μ L.

After discharge, his platelet counts during follow-up appointments at the clinic ranged from 20,000/ μ L to 30,000/ μ L 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.

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