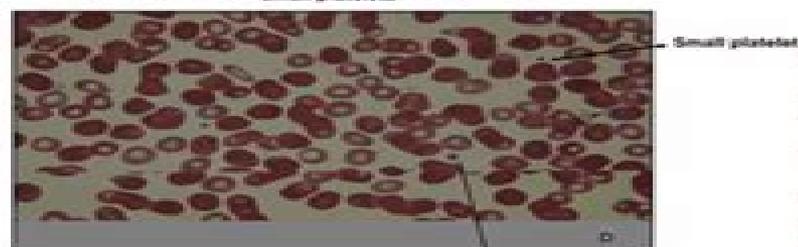




Peripheral smear showing thrombocytopenia and small platelets



Normal sized platelet

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.

Funding: No funding sources

Conflict of interest: None declared

Ethical approval: Not required

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Victor A. McKusick



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