Generalised eczema: a diagnostic clue to Wiskott-Aldrich syndrome

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DESCRIPTION

A 2-year-old boy presented with history of generalised itchy eczematous rashes for one year and loose stool containing blood stains for 10 days. There was a history of skin abscess (left cervical region) at 15 months of age requiring incision and drainage and pneumonia at 18 months of age requiring hospitalisation for 10 days. He was a sixth-born child from a non-consanguineous marriage, there was no history of similar complaints in family members.

On examination, the child was conscious, irritable, however, his vital parameters were within the normal range (Heart rate ; 126/min, Respirtory ate; 26/min, SpO2; 100% in room air, Blood Pressure: 50th centile). He had failure to thrive (weight for length; −4.34 Z score, length for age: −2.66 Z score midarm circumference: 11 cm, head circumference; - 3.12 Z score). He had generalised pruritic eczematous rashes all over the body (figures 1 and 2). Rash was more marked on the trunk with some petechiae, secondary skin thickening, keratinisation and scarring (figure 1). There was excoriation over genitalia (figure 2A), and was associated with bruise over the extremity (figure 2C). Because of recurrent and multisite infection, a possibility of primary immunodeficiency was considered. The haemograms revealed thrombocytopenia with platelet count of 6 x 10^9/L (normal range: 150–450 10^9/L). In immunoglobulin (IG) profile, IgE was >3000 IU/mL (normal range: 20–200 IU/mL) and IgA was 255 mg/dL (normal range (1–3 year): 17–203 mg/dL) were elevated, while IgG and IgM were within normal range. In view of generalised skin rash, thrombocytopenia, blood-stained stool and clinical evidence of immunodeficiency a possibility of Wiskott-Aldrich Syndrome (WAS) was considered. Flow cytometry showed reduced expression of WAS protein (WASp), targeted exon sequencing did not reveal any mutation. However, whole exon sequencing was not done due to financial constraints.

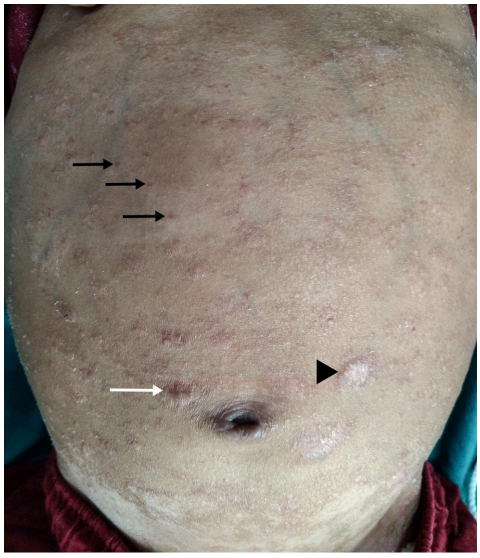

Figure 1 GeneralisedItchy maculopapular rash on the trunk with some petechiae (black arrow), keratinisation (white arrow) and scarring (arrowhead).

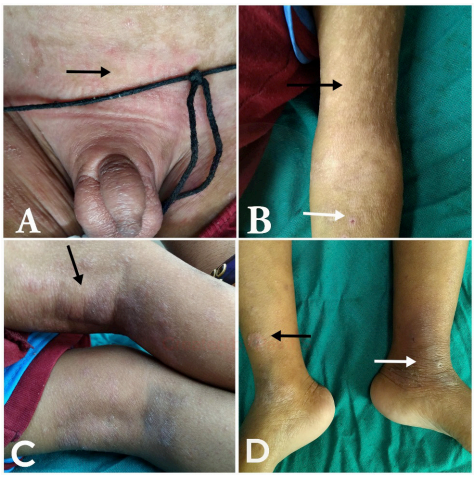

Figure 2 Skin lesion on different parts of the body. (A) Genitalia: diffuse macule with an area of excoriation (black arrow), (B) flexural surface of the forearm; Itchy papular lesion with an area of hypopigmentation (black arrow) and scarring (white arrow), (C) flexural surface of the knee: diffuse maculopapular with an area of bruises (black arrow) and (D) ankle: papular lesion with excoriation (black arrow) and scarring (white arrow).

WAS is an X-linked recessive disorder caused by mutations in the WAS gene which expressed exclusively in haematopoietic cells. WASp is a multidomain protein that exists in complex with several partners that play important roles in its function. Mutations in the WAS gene have various effects on the level of WASp, which, in turn, correlates with the severity of the disease.[1] WAS is characterised by a triad of eczema, thrombocytopenia, and recurrent pyogenic infections.[2] The vast majority of patients had evidence of thrombocytopenia and susceptibility to infection prior to diagnosis, but only 5% of patients present with infections as their only clinical symptom. The cutaneous manifestation in WAS is myriad. Eczema is the most frequent cutaneous manifestation of WAS (71%), followed by petechiae and/or ecchymosis (58%) and cutaneous infections (17%).[2 3] The clinical features of the eczema is

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indistinguishable from atopic or nutritional dermatitis.[3] The severity of eczema may vary and may developed bacterial superinfection in 71% of the cases in the first year of life.[2] The specific treatment of eczema in WAS is not well defined, however several treatment have been attempted including topical emollient and steroids. Topical tacrolimus is an option as a steroid-sparing agent.[4] In it's most severe form, eczema may be resistant to therapy.[4] Intravenous IG is given primary for the severe infection which may also provide transient benefit in skin symptoms.[5]

In the index case, topical emollients and topical steroids were administered without much benefit. Then, we have given intravenous IG @ 600 mg/kg, following that there was a marked improvement in the cutaneous lesions as well as general condition of the child. The child was discharged on antibiotic prophylaxis, and kept on regular follow-up. The parents were counselled regarding its prognosis and available treatment option including haematopoietic stem cell transplantation (HSCT).

Learning points

- A child with generalised eczema with bleeding manifestations with severe, recurrent infections not limited to skin should raise the suspicion of Wiskott-Aldrich syndrome.
- The clinical picture of skin manifestations in WAS may be confused with nutritional dermatitis as most children have associated malnutrition.
- Topical steroids often provide improvement in Eczema. Intravenous immunoglobulin may be needed in severe infection. However, HSCT remain gold standard for the treatment.

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