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Case Presentation

Early-Onset Systemic Autoimmune Disease Presenting with Rash, Edema, And Neurological Involvement: A Case of Monogenic SLE, An Eye Opener

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ABSTRACT

Background: Early-onset systemic inflammatory disorders in children are often challenging to diagnose due to overlapping features of infectious, autoimmune, vasculitis, and metabolic conditions. Rare forms of autoimmune diseases, particularly systemic lupus erythematosus (SLE), may present in infancy with atypical and multisystem involvement including skin, neurological, and systemic features.

Case Presentation: A 1 year 7 months old first order child presented with progressive ankle swelling progressing to generalized edema, followed by 15 days of moderate-grade continuous fever and a generalized painful rash, predominantly over joints. The child had a history of intermittent seizures for 1 year, not on treatment, and global developmental delay without regression with a family history of SLE in mother. On examination, generalized edema, hepatosplenomegaly, and a tender maculopapular rash were noted. Investigations revealed normal WBC count, elevated CRP and ESR (40 mm/hr), hypertriglyceridemia (268 mg/dL), elevated ferritin (349 ng/mL), and raised LDH (1200 U/L). Liver function tests, albumin, and coagulation profile were normal. Sickling test was negative, microcephaly (HC 42 cm) was present, and ANA was positive.

Conclusion: This case emphasizes the need to consider monogenic autoimmune disorders, particularly early-onset SLE, in infants presenting with multisystem involvement and atypical features. Early recognition and appropriate evaluation are crucial for improving clinical outcome

Keywords: *Monogenic SLE, Autoimmune disorders, rash, vasculitis, edema, ANA.*

INTRODUCTION

Early-onset systemic inflammatory disorders in children often present with overlapping features of infection, autoimmune disease, vasculitis, and metabolic disorders, making diagnosis challenging. The presence of rash, fever, edema, hepatosplenomegaly, and neurological involvement should prompt consideration of multisystem inflammatory and autoimmune etiologies, particularly monogenic autoimmune disorders, which tend to present early in life with atypical features.

CASE PRESENTATION

A 1 year 7 months old first order child, born of a non-consanguineous marriage, presented with a total duration of illness of 15 days. The illness started with swelling over both ankles, which was insidious in onset and gradually progressive, later becoming generalized involving the whole body.

This was followed by moderate-grade continuous fever, without chills, rigors, or any localizing symptoms such as cough, diarrhea, vomiting, or urinary complaints. Around 4–8 days into illness, the child developed a generalized rash, initially

over the trunk and later involving the entire body. The rash was predominantly over joints(fig.1), painful and tender, non-vesicular, with no mucosal involvement or bleeding manifestations(fig.2&3).

There was no history of drug intake prior to rash and no classical features suggestive of SLE such as photosensitivity, oral ulcers, or alopecia.

(Fig.1) involvement of knee joint

(Fig. 2) maculopapular rashes all over the body

(Fig. 3) rashes on back

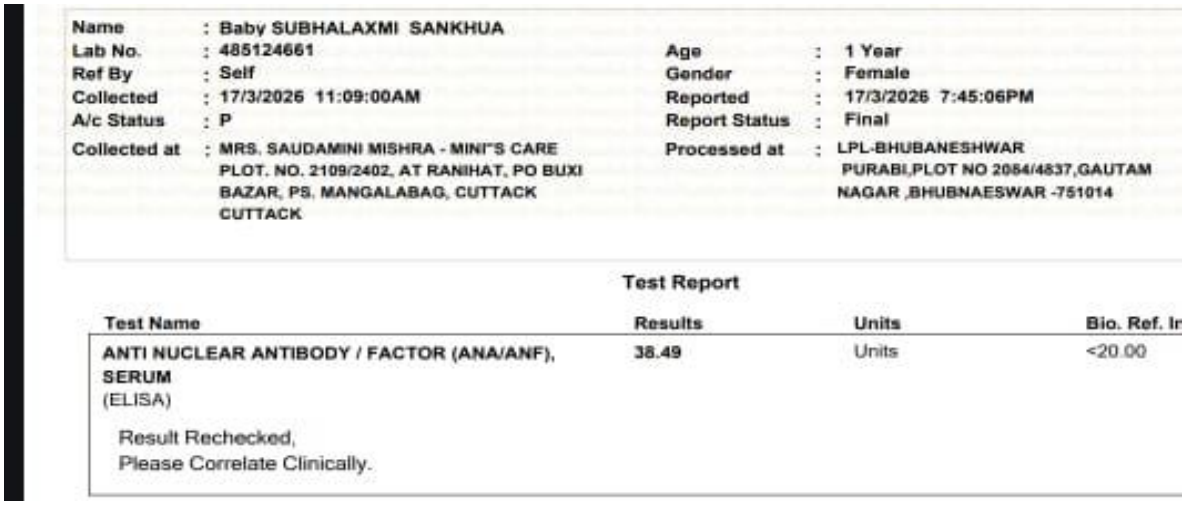
The child had a history of intermittent seizures for the past 1 year, not associated with fever and not on any antiepileptic drugs. Developmental history revealed global developmental delay, with delayed gross motor, fine motor, language, and social milestones, along with poor auditory response, without regression.

Birth history revealed a term LSCS delivery, with birth weight of 1900 gms, immediate cry after birth, and no NICU stay. The child was undernourished (7.5 kg at 19 months) with a significant family history of SLE in mother for which she was under treatment.

On examination, the child was conscious and irritable, with acral edema. Skin examination revealed a generalized maculopapular rash with a reticular (livedo-like) pattern, more prominent over joints and tender on palpation, without vesicles or mucosal lesions. Hepatosplenomegaly was present. CNS examination was significant for developmental delay.

Investigations revealed normal WBC count, elevated CRP and ESR (40 mm/hr), hypertriglyceridemia (268 mg/dL), elevated ferritin (349 ng/mL), and markedly raised LDH (1200 U/L). Liver enzymes, serum albumin, and coagulation profile (PT, APTT, INR) were normal. Sickling test was negative. The child had microcephaly (head circumference 42 cm). ANA was positive (Fig. 3), supporting an autoimmune etiology.

The patient was initiated on oral corticosteroid therapy for control of the underlying inflammatory process. Symptomatic and supportive care was provided. The patient showed initial clinical stability and was advised regular follow-up for further evaluation, monitoring of disease progression, and consideration of additional immunological and genetic workup.



Name	: Baby SUBHALAXMI SANKHUA				
Lab No.	: 485124661		Age	:	1 Year
Ref By	: Self		Gender	:	Female
Collected	: 17/3/2026 11:09:00AM		Reported	:	17/3/2026 7:45:06PM
A/c Status	: P		Report Status	:	Final
Collected at	: MRS. SAUDAMINI MISHRA - MINI"S CARE PLOT. NO. 2109/2402, AT RANIHAT, PO BUXI BAZAR, PS. MANGALABAG, CUTTACK CUTTACK		Processed at	:	LPL-BHUBANESHWAR PURABI,PLOT NO 2084/4837,GAUTAM NAGAR ,BHUBNAESWAR -751014

Test Report

Test Name	Results	Units	Bio. Ref. Ir
ANTI NUCLEAR ANTIBODY / FACTOR (ANA/ANF), SERUM (ELISA)	38.49	Units	<20.00

Result Rechecked,
Please Correlate Clinically.

(Fig. 3), ANA (38.49) POSITIVE REPORT

DISCUSSION

This case represents an early-onset multisystem inflammatory disorder with involvement of skin, CNS, and reticuloendothelial system. Differentials such as sickle cell disease and storage disorders like Gaucher disease were considered and ruled out based on investigations and clinical features. Although some findings suggested hyperinflammatory states, the absence of cytopenias and coagulopathy made them less likely.

The presence of early onset, positive ANA, multisystem involvement, and supportive family history of SLE in mother strongly favors a monogenic autoimmune disorder, most likely monogenic SLE. The patient was initiated on oral corticosteroids, following which clinical stability was achieved, and was advised regular follow-up for further evaluation and long-term management.

Systemic lupus erythematosus (SLE or lupus) is a complex multisystem disease that can affect many organs including the kidneys, skin, joints, lungs, cardiovascular system, central nervous system, and hematopoietic system. SLE is primarily a polygenic disease, with numerous risk variants. Monogenic lupus is a rare form of SLE that begins early in life (usually before age 5) and is caused by mutations in a single gene. It often presents with severe symptoms and is more common in consanguineous families or those with a strong family history of lupus.

Causative defects commonly involve classical complement pathway deficiencies (e.g., C1q, C2, C4), impaired clearance of apoptotic debris, and aberrant nucleic acid sensing pathways, leading to sustained activation of type I interferon signalling. This promotes loss of immune tolerance, chronic inflammation, and early autoantibody production.

Despite its low prevalence, monogenic lupus serves as a model for understanding key pathogenic mechanisms of SLE and enables the development of mechanism-based, targeted therapies.

CONCLUSION

Systemic lupus erythematosus (SLE) predominantly affects adolescent females, reflecting the influence of hormonal and immunological factors on disease expression. However, this case highlights the importance of considering monogenic autoimmune diseases in children presenting with early-onset multisystem inflammatory features.

Early-onset SLE tends to show greater disease activity, increased organ involvement, and a stronger contribution of innate immune dysregulation, particularly involving type I interferon pathways, compared to later-onset disease. Our case highlights the suspicion of monogenic SLE in under 5 children presenting with features of vasculitis.

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