International Journal of Medical and Pharmaceutical Research

Online ISSN-2958-3683 | Print ISSN-2958-3675
Frequency: Bi-Monthly
Available online on: https://ijmpr.in/

Case Report

Turbulent Journey of a Monogenic Lupus- A Case Report

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OPEN ACCESS

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Received: 05-06-2026
Accepted: 25-06-2026
Available online: 10-07-2026

ABSTRACT

Systemic Lupus Erythematosus(SLE) is a chronic autoimmune condition characterised by multisystemic inflammation and presence of circulating autoantibodies directed against self antigens. Though occurs both in children and adults, disproportionately females of reproductive age group are more affected. Monogenic lupus on the other hand has early age of onset along with distinct features including atypical manifestations of underlying disease such as immunodeficiency, immune dysregulation, refractory disease course.

We describe a 7 year old female who presented to us with fever, difficulty in breathing, livedo reticularis rash, generalised swelling of body over a span of 10 days. On Examination pallor, mucositis, generalised lymphadenopathy, hepatosplenomegaly, Previous history was suggestive of three admissions in past. First starting at 3 years of age for fever and mucocutaneous ulceration, resolved with supportive management. Six months apart two more admissions for pneumonia and stroke respectively. Child was on anti seizure medication and anti platelet agent.

Considering early onset of the disease, multisystemic involvement, unexplained fever, mucocutaneous involvement, livedo reticularis rash-provisional diagnosis of early onset lupus(monogenic) was made. Significant investigations like positive ANA, Chest X ray showing cardiomegaly and bilateral pulmonary infiltrates(pneumonia, heart failure), previous MRI, MRA Brain(CNS vasculitis) supported the diagnosis. However Whole Exome Sequencing could not be sent because of limited resources. Treatment with Intravenous pulse methyl prednisolone along with Intravenous antibiotics and other supportive measures. By third day of hospital stay unfortunately, the child required ionotropic and respiratory support, shifted to ICU and succumbed same day.

Despite aggressive treatment the child could not be saved mandating focus on strong suspicion of the disease right from its onset in order to narrow down gap between symptoms and diagnosis. This may help in satisfying outcomes of rare yet fatal disease like monogenic lupus.

Keywords*: SLE, multisystemic, monogenic, lupus, vasculitis, immunodeficiency, rash.*

INTRODUCTION

SLE is a complex autoimmune disease with multisystemic involvement characterised by loss of self tolerance, more of autoantibody formation and immune complex formation. The term 'Monogenic Lupus' is used to describe group of patients with SLE or SLE-like symptoms with a proven underlying pathogenic variant. Those variants are grouped into four major pathways-

1. Complement protein defects
2. Endonuclease gene defects
3. IFN-1 pathways
4. B-& T- cell dysregulation

Combinedly they are called as Interferonopathies. Clinical Features of Monogenic Lupus include-

1. Young age at onset
2. Neurologic features- Spasticity Acute or subacute Dystonia Encephalopathy with seizures & progressive microcephaly Cortical Blindness Developmental Delay Ataxia Psychoses Stroke Demyelinating or multifocal neuropathies
3. Skin features- Chilblains Livedo reticularis Panniculitis Lentigenes Psoriasis Sparse hair, Nail dystrophy
4. Failure to thrive
5. Immune dysregulation-Recurrent fever
6. Hematologic- Dyserythropoesis(Anemia) Thrombocytopenia Malignancy(leukemia, lymphoma)
7. Pulmonary- Interstitial Lung Disease Intraalveolar hemorrhage
8. Vascular – Calcification of aorta and other blood vessels
9. Musculoskeletal- Joint pain, arthritis Contractures and joint retractions
Myositis
10. Ophthalmologic- Glaucoma
11. Kidney- Lupus Nephritis
12. Gastrointestinal- Very Early Onset IBD
13. Dental Anomalies- Retained primary teeth Premature loss of permanent teeth

CASE PRESENTATION

A 7 year old female admitted to our hospital with a ten days history of fever, difficulty in breathing, livedo reticularis rash, generalised swelling of body.

In past, at the age of 3 years the child had first presentation of fever for 2 weeks on and off for 1 week with some facial rash, resolved with some oral medications.

Six months later child presented to another institute with acute onset weakness and 2 episodes of unprovoked seizure- treated in line of Stroke with antiplatelet agent and anti-seizure medication which was continued till this presentation. Eight months later, the child had been admitted to another institute for Pneumonia with a stay for 6 days, discharged. Child was unable to attend school because of disability and also had loss of appetite.

On Examination

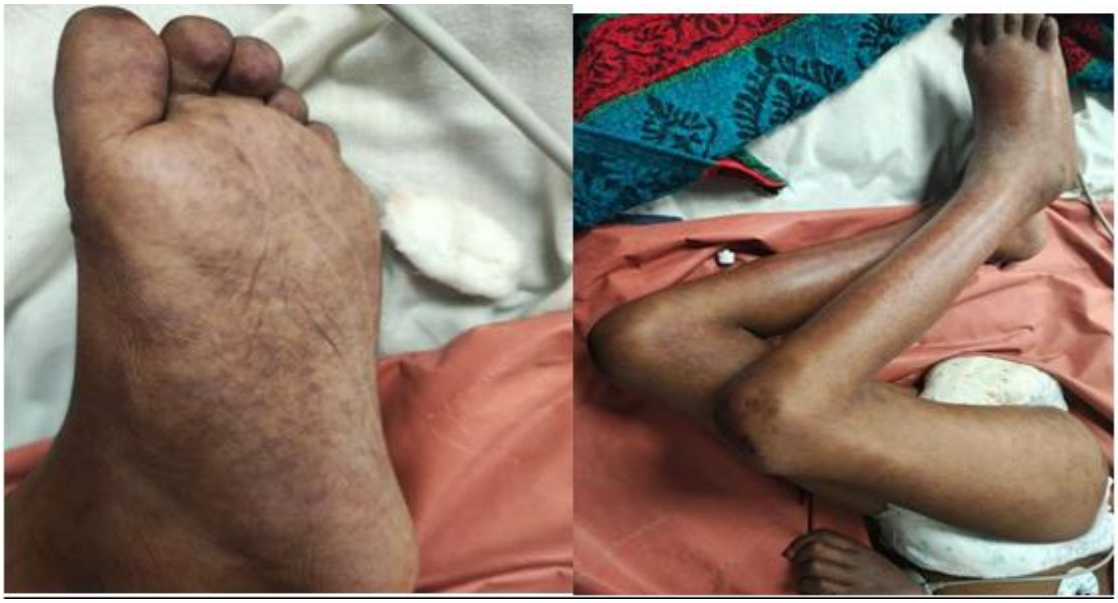

Figure 1 – Chilblain, Figure 2- Livedo Reticularis and joint contractures (spasticity)

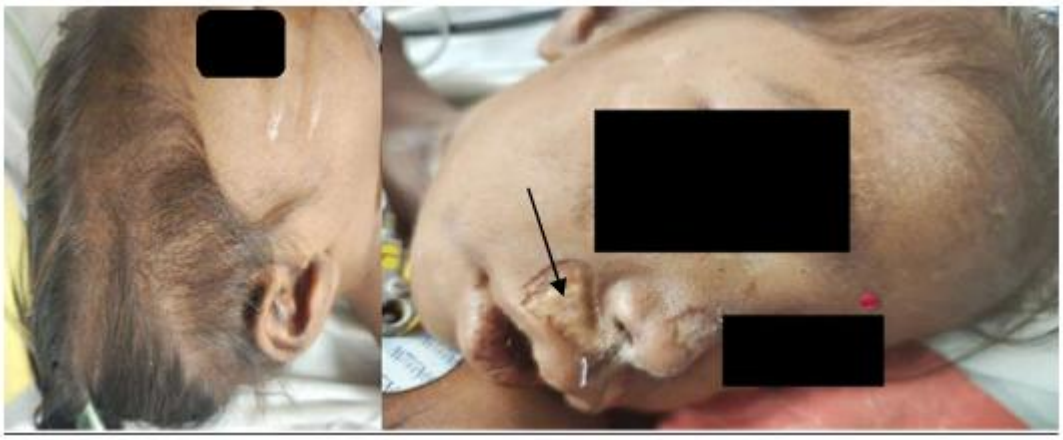

Figure 3- Alopecia **Figure 4- mucocutaneous ulcers**

Generalised wasting present.
Alopecia present. Mucocutanous lesions(oral ulcers, healed discoid lesions over nasolabial region) present. Hyperpigmented skin.

Pallor, Livedo reticularis rash,chilblains, anasarca present.
Febrile, Tachypnea, Tachycardia with hypoxia in room air(92% in room air) while blood pressure maintained.
Higher Mental Function-Alert, oriented, poor attention, memory intact, executive function and abstract thinking diminished.

Spasticity and joint contractures present bilaterally, more on left upper and lower limbs.
Deep tendon reflexes exaggerated while superficial reflex – plantar showed extensor response. Power diminished left>right muscle groups of both upper and lower limbs.

No active convulsion. No meningeal signs. Respiratory – bilateral diffuse crepts present Cardiovascular- muffled heart sounds with no murmur

Abdomen- soft, Hepato splenomegaly(liver span-12cm, spleen-7 cm), slit like umbilicus.
We made a Provisional diagnosis of "Monogenic Lupus with Pneumonia in Congestive Cardiac Failure with CNS vasculitis".

Supporting investigations -

CBC- Hb 7.8 MCV 81, MCH 28.9 MCHC 35.5 TLC 11610 N 73.4 L 19.6 M 5.1 E1.4 TPC 3.5
CRP(Q)- 84mg/dl
ESR-100mm/hr
SGPT/SGOT/Serum Albumin/Total bilirubin-78/58/2/1.2
Serum Urea/Creatinine- 48/1.1
Urine Routine Microscopy- Albumin 1+
Chest X ray – B/L pulmonary patchy infiltrates with cardiomegaly
2D ECHO- LV Dysfunction, Ejection Fraction-38%, mild Pericardial effusion
MRI, MRA Brain- CNS Vasculitis
ANA positive

During Hospital stay, IV methyl prednisolone was started at 20mg/kg/day along with IV Antibiotics. Anti seizure medication was continued. Iv ionotropes were required on day 2 of stay. Shifted to ICU because of worsening hemodynamic instability and mechanical ventilation support. Unfortunately the child succumbed in ICU on third day of hospitalisation.

DISCUSSION
Ubiquotously expressed inflammatory polypeptides induced by viral and microbial nucleic acids are type 1 interferons. Altered regulation of Interferon signalling causes unique clinical syndromes classified as interferonopathies. Characteristic phenotype being recurrent fevers, early onset skin vasculopathy(chilblains, livedo reticularis, panniculitis,

lipodystrophy), ILD with fibrosis, encephalopathic CNS involvement and spasticity.

Our case discussed got fit into almost all domains of monogenic lupus, yet to regret it was too late to modify the disease to hault the fatality.

Thus early suspicion strongly based on the characteristic skin presentation, unexplained fever, multisystemic involvement with a young onset can lead to early diagnosis, appropriate investigations and early start of disease targeted therapy such that the disease takes a modified journey with favourable outcomes.

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