



A RARE CONVERGENCE:

“Multisystem Langerhans Cell Histiocytosis with Sickle Cell Trait in a Pediatric Patient”



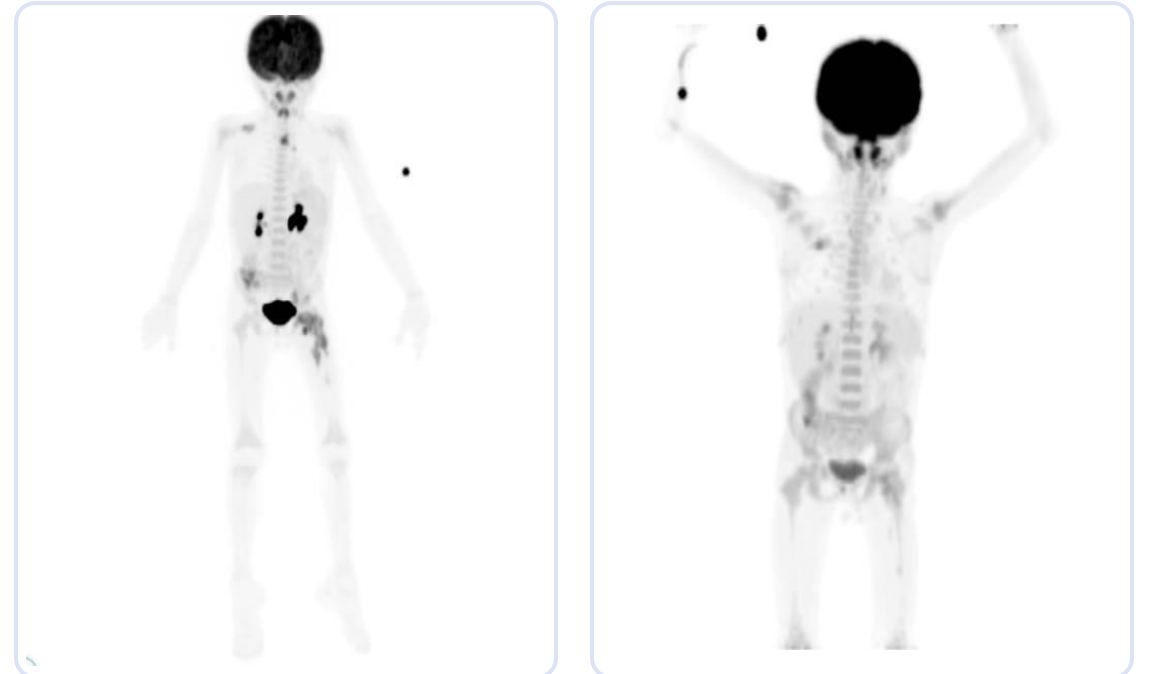
Dr. Zainab Nawab, Dr. Farrukh Zia Tareen, Dr. Zoha Rasool

Correspondence: zainabrashid2100@gmail.com

BACKGROUND

- LCH is a rare, potentially high-risk **myeloid neoplasm** caused by proliferation of pathogenic dendritic cells.
- In TB-endemic regions like Pakistan, LCH closely mimics **tuberculous osteomyelitis**, causing significant diagnostic lag.
- Diagnosis relies on **tissue biopsy**, immunohistochemistry (CD1a, S-100, CD207) and **BRAF V600E** mutation testing.
- Persistent osteolytic lesions unresponsive to antibiotics or anti-tuberculous therapy warrant **early biopsy**.
- Coexisting hemoglobinopathies can add diagnostic noise, further delaying recognition of the underlying neoplasm.

IMAGING



Whole-body 18F-FDG PET Maximum Intensity Projection (MIP). **Fig. 1 (left):** posterior view, upper extremities elevated. **Fig. 2 (right):** anterior view, upper extremities lowered.

CASE HISTORY

PRESENTATION A **6-year-old boy** with a 2-year history of recurrent scalp swelling with pus discharge, refractory to antibiotics and 3 months of anti-tuberculous therapy.

WORKUP Persistent anemia workup via HPLC revealed coexisting **Sickle Cell Trait (HbS 37.1%)**. Skull X-ray showed lytic lesions; scalp biopsy established the diagnosis.

HISTOLOGY Sheets of mononuclear cells with grooved coffee-bean nuclei, eosinophils and multinucleated giant cells. IHC positive for **CD1a, S-100 and CD163**; GeneXpert and AFB smears negative.

DIAGNOSIS PCR confirmed **BRAF V600E** mutation, establishing **Multisystem LCH (Low Risk)**.

BASELINE PET-CT Multifocal FDG-avid lesions in the frontal bone, mandible, scapula, iliac bone and pelvic lymph nodes.

TREATMENT 6-week **LCH-IV induction**: weekly Vinblastine, daily Prednisolone, Bisphosphonates.

RESPONSE Follow-up PET-CT showed complete nodal resolution and decreased skeletal metabolic activity (SUVmax 6.0 to 1.5–1.7), confirming **partial response**. A second 6-week reinduction block was initiated.

CONCLUSION

- In TB-endemic regions, persistent osteolytic lesions unresponsive to antibiotics or ATT require **prompt tissue biopsy** to exclude LCH.
- Coexisting hemoglobinopathies may mask underlying neoplasia, delaying tissue diagnosis.
- Biopsy with IHC and molecular identification of **BRAF V600E** confirms the diagnosis, ensuring timely initiation of protocolised multisystem therapy.

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