Systemic mastocytosis: A rare differential diagnosis to be considered in pediatric patients presenting with cutaneous mastocytosis and seizures

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Sir,

Mastocytosis is a rare disorder characterized by clonal proliferation and accumulation of mast cells in different organs, leading to varying clinical pictures. Cutaneous mastocytosis or urticaria pigmentosa involves only skin and affects pediatric patients. Systemic mastocytosis is seen mainly in adults and affects multiple organs including the brain [1].

We are reporting the case of a 3 year-old-girl who presented with four episodes of seizures over a period of 3 months. She had brownish macules and infiltrated plaques with severe itching, redness, and blistering over the face, trunk, and legs for 3 months of age. A family history of seizures was present. On examination Darier's sign and antinuclear antibody were positive. The seizure was diagnosed as early-onset occipital lobe epilepsy. Magnetic resonance imaging brain showed focal abnormal sulcation with cortical thickening in the left postero-inferior parietal region adjacent to the parieto-occipital sulcus. ASL perfusion map showed subtle asymmetric perfusion in the corresponding region. An intermittent spike and sharp wave discharge over the left frontocentral region was seen in electroencephalogram.

Skin lesions were biopsied and microscopy revealed reticular dermis with loose sheet of mast cells with oval or round elongated nuclei and granular cytoplasm dissecting dermal collagen bundles in deeper dermis suggestive of cutaneous mastocytosis (Fig. 1). In view of cutaneous mastocytosis with seizure, a differential diagnosis of systemic mastocytosis was considered. She was managed with lorazepam 1.2 mg intravenously, sodium valproate 240 mg slow intravenous infusion over 1 h followed by 100 mg intravenously twice daily, and phenytoin 30 mg intravenously twice daily for control of seizures along with ketotifen for cutaneous lesions. After initiation of these treatment modalities, her condition improved and there were no further episodes of convulsion. Further investigations for diagnostic confirmation of systemic mastocytosis with bone marrow and immunohistochemistry studies are awaited.

Cutaneous mastocytosis usually has a benign presentation. Systemic mastocytosis, an aggressive form of the disorder, may affect the bone marrow, liver, spleen, lymph nodes, and rarely the brain. The most frequent symptoms include pruritis, flushing, itching, diarrhea, and anaphylaxis [2-4]. The clinical manifestations of mastocytosis result from mast cell-derived mediators and, less frequently, from destructive infiltration of mast cells. Mast cells are specifically located in the dura mater or meninges of the spinal cord and brain. Increased blood-brain barrier permeability can lead to mastocytes crossing into the central nervous system and is associated with higher levels of neuroinflammation and brain dysfunction [5].

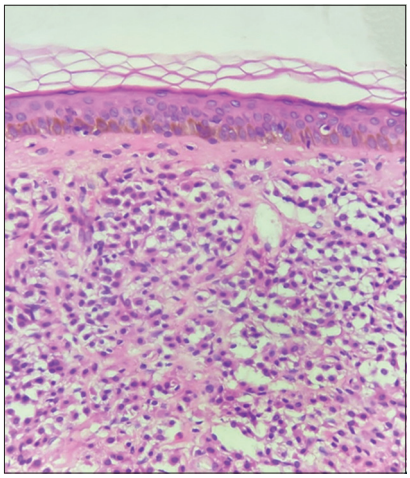

Figure 1: High-power microscopy of skin lesions showing reticular dermis with loose sheets of mast cells

The diagnosis is based on the World Health Organization criteria, which includes tryptase level, histopathological and immunophenotypic evaluation of mast cells, and molecular analysis [1]. A somatic KIT mutation, most commonly D816V, is usually detectable. Mastocytosis comprises seven different categories that range from indolent forms like cutaneous and indolent systemic mastocytosis to progressive forms, such as aggressive systemic mastocytosis and mast cell leukemia. Although prognosis is good in patients with mild forms of the disease, patients with advanced categories have a poor prognosis [1,4]. Ideally, patients with systemic mastocytosis should undergo next-generation sequencing to identify potential

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genetic carriers. The important differential diagnosis in patients with cutaneous mastocytosis and seizures includes systemic mastocytosis with cutaneous involvement, neurocutaneous syndrome, and mast cell activation syndrome.

It is concluded that when a child with cutaneous mastocytosis presents with seizures, a thorough evaluation to rule out systemic effects of mastocytosis including bone marrow biopsy, radiological imaging, and electroencephalography is vital.

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