Hand–Schuller–Christian disease presenting with a classical triad and oral manifestations: A rare case report

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Abstract

Hand–Schuller–Christian disease is a multifocal disease that characteristically involves the bone and extraskeletal involvement of the reticuloendothelial system. The most common age group affected is children between 1 and 5 years old. It is a disseminated form of Langerhans cell histiocytosis with a typical triad of exophthalmos, diabetes insipidus, and lytic bone lesions, although seen in only half of the patients. We present such an uncommon case presenting with the classical triad along with oral manifestation such as floating teeth and hepatitis with typical radiological findings.

Key words: Hand–Schuller–Christian disease, Triad, Floating teeth

Langerhans cell histiocytosis (LCH) is a condition characterized by the proliferation of dendritic mononuclear cells that infiltrate the organs. Rarely seen in adults, these most commonly involve infants and children. Hand–Schüller–Christian (HSC) disease is one of three clinical syndromes noted in LCH. The other two include eosinophilic granuloma and Letterer–Siwe disease [1]. Eosinophilic granuloma presents with single or multiple bone lesions without any visceral involvement. Letterer–Siwe disease is disseminated form of LCH which involves multiple organs and carries the worst prognosis among the three. HSC is a chronic disseminated form of LCH presents with a classic triad of skull lesions, exophthalmos, and diabetes insipidus. However, the classical triad of HSC is seen in only 30% of the patients [2]. We report such a case of HSC which present with the classical triad along with oral manifestations.

Case Report

A 3-year-old male child was referred to the radiology department of our hospital for the evaluation of the abdominal distension. The mother complained of gradually increasing abdominal distension for the past 6 months. The previous two visits to the nearby clinic failed to find any diagnosis. We also noticed macrocephaly which on further questioning, mother told us that the head has been enlarging for the past 1 year. The patient’s mother also brought our attention to a painful swelling over the scalp which was also slowly increasing for the past 1 year. The mother also complained of excessive water intake by the child resulting in polyuria and polydipsia.

On examination, the vitals were stable. Local examination revealed uniform distension of the abdomen. No obvious focal abdominal swelling was visualized. The liver was palpable 5 cm below the right costal margin. The spleen was mildly palpable below the left costal margin. The scalp swelling was present in the right parietal region. It was 2 cm in size with a smooth margin, firm in consistency, and mildly tender. No obvious swelling or tenderness noted over the maxilla or the mandibular region.

Ultrasonography (USG) of the abdomen revealed marked hepatomegaly with periportal cuffing which was suggestive of changes of hepatitis and an overdistended bladder. Spleen was mildly enlarged. Curious about the macrocephaly, we also performed USG of the head through the temporal window. No obvious hydrocephalus was noted. The radiograph of the skull and face revealed multiple lytic lesions with beveled edges and mandibular floating teeth (Fig. 1). These imaging findings were suspicious for HSC. Hence, a computed tomography (CT) scan of the head was performed. CT scan further confirmed our findings (Fig. 2). A 3D volume-rendered images demonstrating the floating teeth (Fig. 3). Magnetic resonance imaging (MRI) scan of the sellar region revealed a large enhancing nodular mass lesion in the sellar region with an absent posterior pituitary bright spot (Fig. 4). MRI findings were consistent with diabetes insipidus.

In view of the classical triad, the patient was diagnosed with HSC disease which was further confirmed on a simple punch biopsy of the skin. Due to multisystem involvement, the patient was started on combination therapy of prednisone and cytarabine. At 6-week follow-up visit, the abdominal distension and scalp swelling had significantly decreased. The patient is on regular follow-up.
DISCUSSION

Hand–Schuller–Christian (HSC) disease is an idiopathic condition where the non-neoplastic proliferation of histiocytes occurs. HSC is a chronic disseminated form of Langerhans cell histiocytosis. The classical triad of HSC includes exophthalmos, diabetes insipidus, and lytic bone lesions. However, only one-third of the patients present with the classical triad. Infants and children are most commonly involved with rare involvement in adults [1-3].

The most commonly involved structure is the skull, followed by the skin, lymph node, and lung. Diabetes insipidus is seen in half of the patient with HSC [3]. Skin involvement in the form of erythematous scaly rash is seen in 30% of patients. Other less common features include poor sexual development, retarded growth, central and perihilar pulmonary involvement, extensive interstitial fibrosis, cor pulmonale, hepatosplenomegaly, and thyroid involvement [4].

HSC most commonly involves membranous bones. In the case of long bones, it involves diploe with epiphyseal lesions being unusual. The bony lesions are usually multiple and appear as punched-out lytic lesions with beveled edges. These lesions are commonly found in the skull, mandible, ribs, scapula, and pelvis. Some of these lesions may coalesce together to give a geographic appearance [5-7]. A subgroup of patients may present with oral lesions as their initial manifestation ranging from foul-smelling breath, gum infection, and altered taste to alveolar involvement. The severe alveolar involvement may result in what is referred to as “floating teeth” appearance. Spinal involvement may present as vertebra plana resulting from the gradual collapse of the vertebral body.

CT and MRI may be useful to define the extent of organ involvement. Contrast MRI is useful for the evaluation of the sellar and suprasellar involvement. The posterior pituitary gland involvement leads to diabetes insipidus. The differentials for sellar and suprasellar lesions include granulomatous lesions and hypothalamic gliomas [8]. Nuclear bone scans falsely negative in many cases [9,10].

The differential diagnosis includes skull lesions that include osteomyelitis and epidermoid cyst. The differentials for an aggressive form of long bone involvement include round cell lesions, Ewing’s sarcoma, osteosarcoma, leukemia, acute osteomyelitis, and less aggressive form which include simple cysts, fibrous dysplasia, and chronic osteomyelitis. The spine involvement (vertebra plana) includes leukemia, metastatic neuroblastoma, and idiopathic osteonecrosis [6].
CONCLUSION

Radiologists must be aware of the imaging spectrum of HSC which helps to detect systemic involvement. Early detection and treatment can avoid serious complications and morbidity to the patient.

REFERENCES


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