Case Reports

An unusual case of histiocytic sarcoma

Introduction

Histiocytic sarcoma is an uncommon neoplasm with only a few reported cases till date[1,-3] Previously some lymphoid malignancies were misclassified as histiocytic lymphomas due to overlapping features and lack of immuno-histochemical evidence.[1,2,4] Here, we discuss a case report of this rare neoplasm in the terminal ileum of a female patient. The morphologic and immunohistochemical features of the neoplasm are described in detail, and its differential diagnoses excluded.

Case report

A 59-year old diabetic female presented with complaint of abdominal pain. CT abdomen showed a mass in the terminal ileum **(Figure 1).** PET scan was done and no other lesion was identified. Surgical resection was performed.

Result

On pathologic examination, the surgical specimen consisted of a segment of small bowel. In the center of the specimen a polypoid well-defined tumor mass projecting into the lumen was seen. The tumor measured 6 cm × 5 cm. The overlying mucosa was intact. The mass extended up to the serosal surface. The cut surface was tan pink homogenous and soft. **(Figure 2)**

Microscopically, the tumor was composed of sheets of large epithelioid cells with abundant eosinophilic cytoplasm, oval to irregular nuclei, vesicular chromatin, and large nucleoli. Bi-

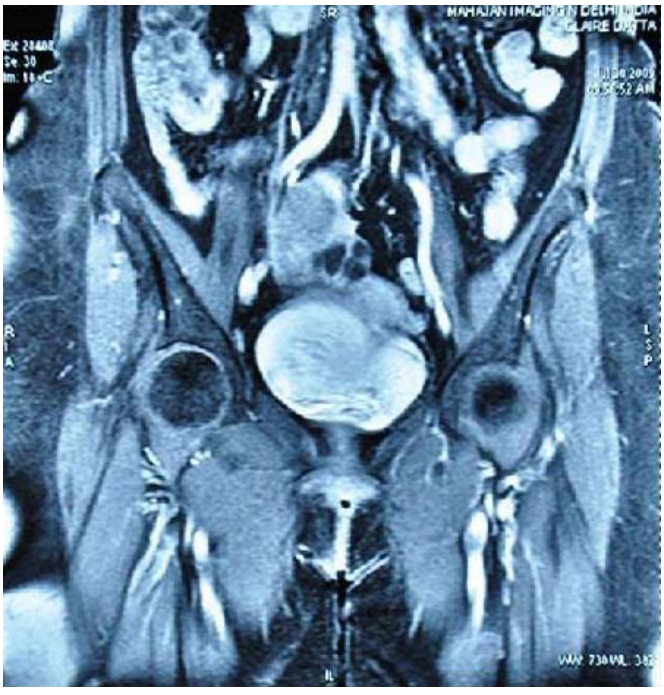

Figure 1: CT scan shows a mass measuring 6x5cm in terminal ileum

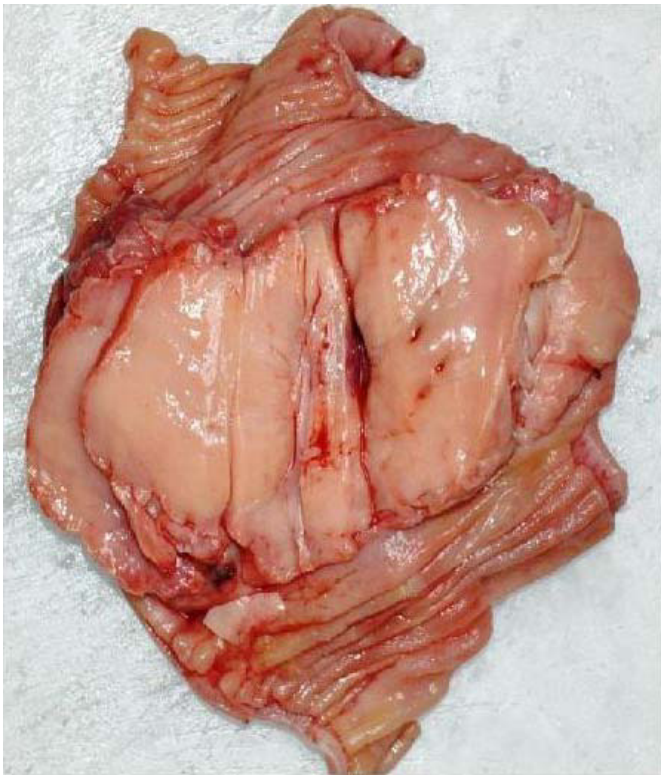

Figure 2: Shows a well defined polypoidal tumor mass projection into lumen of small bowel cut section tan pink, homogenous and soft

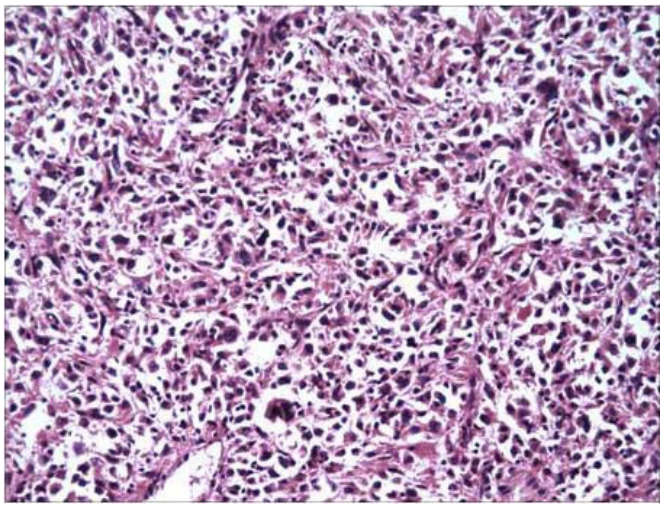

Figure 3: Tumor is composed of sheets of large epithelioid cells with abundant eosinophilic cytoplasm, oval to irregular nuclei, vascular chromatin and large nucleoli (H&E, x40)

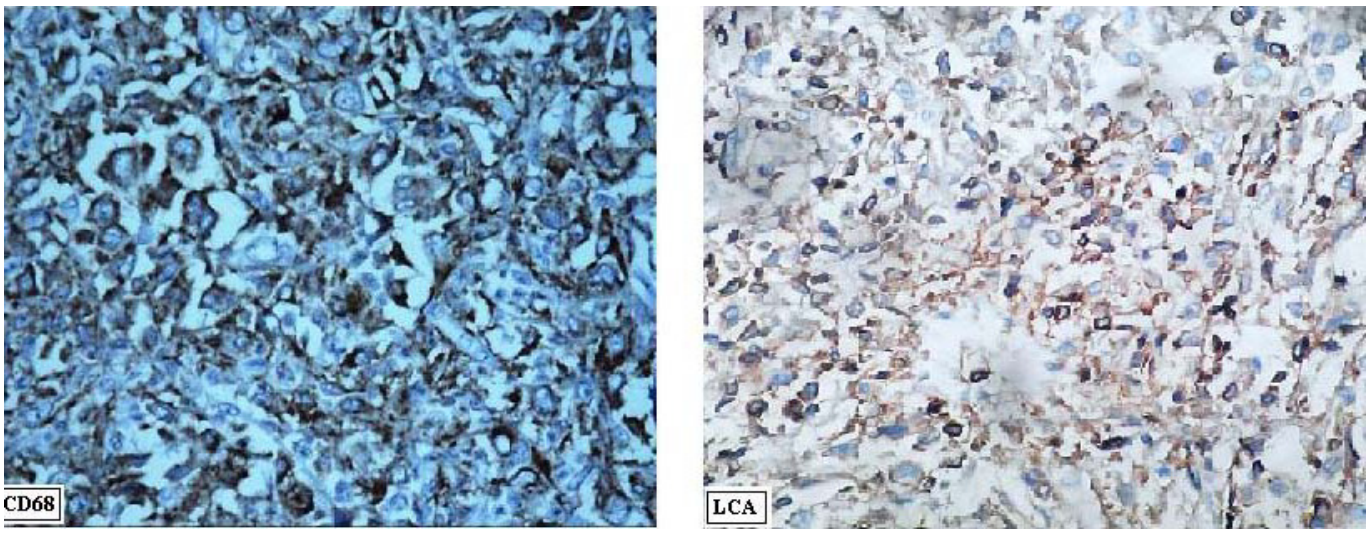

Figure 4: Shows large tumor cells positivity with CD68 immunostains (IHC,x40) LCA immunostain (IHC, x60)

nucleated cells were common. Mitoses ranged from 3-4 per 10 HPF. These cells infiltrated the muscularis propria as well as the mucosa. There was intense inflammatory infiltrate in the background**. (Figure 3)** On immunohistochemical examination the large tumor cells were LCA and CD68 positive. **(Figure 4)** The tumor cells were negative for cytokeratins, CD34, CD3, CD20, ALK-1, CD21, myeloperoxidase, HMB-45, c-kit and Desmin.

Discussion

With only a limited number of cases reported so far, Histiocytic sarcoma (HS) is a relatively new entity. Although it spans a wide age range most cases are documented in adulthood.[5] Gastro-intestinal involvement caters to only one third of the total cases.[1,2,3,5,6] It manifests in the form of abdominal mass, pain abdomen, intestinal obstruction, lower gastrointestinal bleeding or weight loss.[7] Its etiology is not known.

At the present time, the World Health Organization defines HS as a malignancy with morphologic and immunophenotypic features resembling those of mature tissue histiocytes.[3,8,9] Features distinguishing malignancy from a benign histiocytic lesion include presence of malignant cytologic features with evidence of tissue invasion.[3] The latter features were seen in our case. The diagnosis of HS however relies predominantly on the verification of histiocytic lineage and the exclusion of other, poorly differentiated, large cell malignancies (i.e. lymphoma, carcinoma and melanoma) by way of extensive immunophenotypic investigation. Another strong differential includes leukemia of monocytic origin. Nonetheless, absence of a leukemic phase with circulating tumor cells supports the diagnosis of HS over the latter.

Our immunohistochemical analysis included stains to confidently exclude melanoma (i.e. melan A, HMB-45), carcinoma (i.e. cytokeratin), lymphoma (CD3, CD15, CD20, CD30, CD43, CD79a, ALK1) and accessory/dendritic cell tumors (i.e. CD1a, CD21). Since the mature tissue histiocyte is believed to be the benign counterpart of HS, CD45 (LCA) immunoreactivity is a requirement to establish hematopoietic origin.[3] It also allowed differentiation from morphologically similar soft-tissue neoplasms. While CD45 immunoreactivity may be an absolute requirement, with the aid of additional supportive studies such as electron microscopy, differentiation from malignant fibrous histiocytoma (undifferentiated sarcoma) may be extremely difficult. These latter tumors are believed to arise from undifferentiated mesenchymal cells rather than histiocytes, and typically show pleomorphic and storiform growth patterns.[3] Despite these subtle morphologic and immunophenotypic differences, clinical history may be the only means of differentiating HS from a soft-tissue malignancy. Extranodal presentation is not that frequent and the clinical course is generally aggressive.[1-3] On the whole, unfortunately, persistent clinical and histopathologic disparities in these studies, in addition to the overall rarity of these neoplasms have continued to obscure this diagnosis and have prevented a full appreciation of their clinical behavior.

Treatment options for HS include surgery, chemotherapy and radiotherapy. Surgical resection with wide margins is the gold standard of treatment.[8] The morphologic and immunophenotypic features of HS are relatively uniform, however, an extensive panel of immunohistochemical markers is necessary to adequately exclude more common, poorly

differentiated malignancies.

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Acute intermittent porphyria: a missed diagnosis in pre-pubertal children with recurrent abdominal pain

Introduction

Acute intermittent porphyria (AIP) is the most common hepatic porphyria. However, diagnosis of AIP is often delayed as its manifestations are variable and non-specific. AIP is a disorder of the third or fourth decade, it rarely manifests before puberty. Here we report two cases of AIP who presented in pre pubertal age.

Case 1

A 9-year old boy presented to the outpatient services with a 2-year history of recurrent abdominal pain. The pain was periumblical, episodic, moderate in intensity, once every 2-3 months, lasting for a few hours. There was no history of abdominal distension, vomiting or constipation. Physical examination was unremarkable with normal growth parameters. The child was evaluated elsewhere outside our hospital with complete blood counts, serum amylase, x–ray abdomen, urine and stool examination, abdominal ultrasound and a CECT abdomen. All these investigations were reported normal. The child was suspected to have functional abdominal pain possibly abdominal migraine as the episodes were intense, paroxysmal and intermittent with pain-free periods between the episodes. After one more visit to our outpatient department a month later,the child was lost to follow-up.

Six months later he presented to the emergency with seizures. At presentation he was in a post–ictal state, with a Glasgow coma score of9/15(E3M4V2), heart rate of 134/minute and BP of 174/110 mm Hg. There was no focal deficit and meningeal signs were absent. Fundus examination was normal. The abdomen was slightly distended with sluggish bowel sounds. There was no organomegaly, tenderness or guarding and liver dullness was preserved. Sexual maturity rating was pre–pubertal and there were no other findings on general physical and systemic examination.

Laboratory investigations showed hyponatremia (serum sodium 116 meq/L) and hypokalemia (serum potassium 3.3 meq/L).Serum calcium, magnesium and blood sugars were normal. A CT head and CSF examination done elsewhere were unremarkable. Plain X–ray abdomen (erect) showed dilated bowel loops (**Figure 1**). Urine for porphobilinogen was