

DIAGNOSIS:

The overall features are suggestive of bone marrow infiltration by Ewing sarcoma spectrum of neoplastic disease.

Advice:

EWSR1 gene rearrangement for confirmation of the diagnosis.

DISCUSSION:

Ewing sarcoma is a part of a spectrum of neoplastic diseases known as the ES family of tumors (EFT), which also includes extrasosseous ES (EES), PNET, malignant small-cell tumors of the thoracopulmonary region (Askin tumor) and atypical ES.

Ewing sarcoma tends to arise in the diaphysis or metaphyseal –diaphyseal portion of long bones. The pelvis, ribs are also common locations [2].

The skull, vertebrae, scapula and short tubular bones of hand and feet are rarely involved. It is extremely rare to find lytic lesions in spine (8%) and skull(4%)[3,4]

Most cases of Ewing sarcoma show uniform small round cells with round nuclei containing finely stippled chromatin and inconspicuous nucleoli, scant clear or eosinophilic cytoplasm, and indistinct cytoplasmic membranes [2].

95% of Ewing sarcoma express high levels of a cell surface glycoprotein encoded by CD99 (MIC2X) gene which show diffuse membranous positivity on immunohistochemistry which is a relevant diagnostic marker[5].

Ewing sarcoma are associated with structural rearrangements that generate FET-ETS fusion genes.

FLI1 and ERG are often expressed in the cases with the corresponding gene fusions.[6]

More than 85% of Ewing's sarcomas are defined by the t(11;22)(q24;q12) translocation that fuses the EWSR1 gene on chromosome 22 with the FLI1 gene on chromosome 11.[7]

The same was advised in the present case for confirmation of the diagnosis.