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Unveiling the diagnosis of rhabdomyosarcoma on bone marrow biopsy without an obvious primary

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Quick Response Code:

A 13-year-old girl presented with one-month history of fever, dyspnoea, pallor, multiple petechiae over bilateral lower limbs and reduced air entry in left hemithorax. CT scan chest showed multiple pleural nodules, massive left-sided effusion with multiple osteolytic lesions in the overlying ribs. Bone marrow biopsy showed infiltration by abnormal small cells with hyperchromatic nuclei forming vague nodules at places. On immunohistochemistry (IHC), these abnormal cells mimicking blasts were negative for acute leukemia markers (LCA, TdT, CD34, CD117, PAX5, CD3, MPO, CD99) and were strongly positive for Desmin, Myogenin, Myo D1, Vimentin, CD56 and negative for NSE, Synaptophysin, Chromogranin, PanCK, WT1 [Figure]. IHC findings

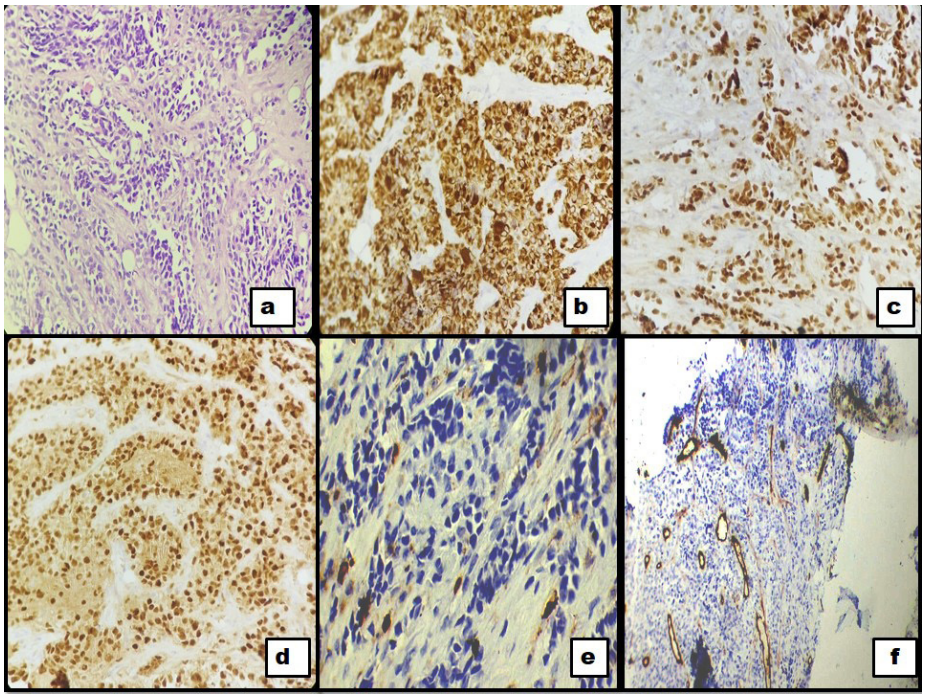

Figure 1: A case of rhabdomyosarcoma with bone marrow infiltration a) H&E stained section of bone marrow biopsy (40X) with infiltration by abnormal small cells with hyperchromatic nuclei. b-d) Desmin (40X), MyoD1(40X), Myogenin (40X) shows diffuse positivity in these cells. e and f) LCA (40X) and CD34 (10X) are negative in the abnormal round cells.

suggested a diagnosis of infiltration of bone marrow by soft tissue sarcoma (likely to be rhabdomyosarcoma) was suggested.

Rhabdomyosarcoma is the most common soft tissue sarcoma of adolescent and childhood; however, bone marrow involvement as presenting feature without an obvious primary is an extremely rare phenomenon[1].

Declaration of patient consent

Patient's consent not required as patients identity is not disclosed or compromised.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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