

Metastatic rhabdomyosarcoma initially diagnosed on the bone marrow



A 15-year-old boy presented with a rapid-onset right-sided facial mass, thrombocytopenia $(114 \times 10^{9}/L)$, and elevated serum lactate dehydrogenase. A computed tomography scan demonstrated a maxillary sinus and nasopharyngeal mass with extensive cervical lymphadenopathy. Bone marrow (BM) biopsy was performed shortly prior to lymph node resection due to high suspicion of lymphoma. BM aspirates showed numerous discohesive cells with scant-moderate vacuolated basophilic cytoplasm, dispersed nuclear chromatin, and prominent nucleoli (panel A; original magnification ×1000, Wright-Giemsa stain). BM biopsy showed interstitial and diffuse mononuclear cell infiltrates (panel B; original magnification ×400, hematoxylin and eosin stain). Flow cytometry showed a discrete CD56⁺ cell population, negative for lymphoid/myeloid markers (panel D). An extensive panel of immunohistochemical stains showed myogenin (panel C; original magnification ×400) and desmin-positive infiltrates, diagnostic of metastatic rhabdomyosarcoma. Subsequent histopathological workup of the resected lymph node, and positive *FOXO1A* (*FKHR*) gene rearrangement [t(2;13)(q33;q14)] by fluorescence in situ hybridization (dual-color, break-apart rearrangement probe; panel E) confirmed a solid variant of alveolar rhabdomyosarcoma. The patient underwent chemotherapy and radiation, but unfortunately died 2 years after diagnosis due to widespread metastases.

Rapid diagnosis was possible on the BM specimen using a combination of flow immunophenotyping and immunohistochemical workup, despite the absence of any distinctive morphologic findings. The case highlights the importance of a comprehensive BM workup using all modalities.



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