Abstract

The clinical, histologic and cytogenetic features of a patient with the alveolar subtype of rhabdomyosarcoma (RMS) were investigated. The patient presented with a widely disseminated tumour including bone marrow involvement, and was a diagnostic dilemma. The presence of translocation (2;13)(q37;q14), which is strongly associated with alveolar RMS helped make the diagnosis. A review of other published cases confirms the strong association of (2;13) with alveolar RMS. The importance of considering RMS as a differential diagnosis in patients presenting with disseminated tumour as the only finding is stressed. This case also shows how cytogenetic investigation of similar patients may provide a diagnosis.