Background Small round cell tumours, particularly rhabdomyosarcoma (RMS), may infiltrate bone marrow (BM), mimicking acute leukaemia – both clinically and in morphological assessment of myelogram.

Aim To analyse diagnostic and therapeutic dilemmas in children with RMS masquerading as acute leukaemia.

Methods A retrospective analysis of medical charts of 14-year-old male and 15-year-old female admitted to Department of Paediatrics, Haematology and Oncology, Medical University of Gdansk, Poland in 2007 and 2013 and literature review.

Results Both patients were referred suspected of acute leukaemia presenting with weakness, pallor, bone pains, and enlarged peripheral lymph nodes (LN). Skin bruising, petechiae and wound bleedings were progressing. Laboratory tests showed anaemia, thrombocytopenia and features of acute DIC and ATLS. BM aspiration revealed blast cells suggestive for leukaemia. Flow cytometry failed to display lymphoid or myeloid antigens. Aspirational LN biopsy revealed small round blue cells, suggesting AML, non-Hodgkin’s lymphoma or Ewing’s sarcoma. Modified chemotherapy in all these malignancies was introduced to alleviate DIC-associated haemorrhages and enable LN resection. Finally the diagnoses of embryonal RMS (male) and alveolar RMS (female) were made. Administration of proper chemotherapy for metastatic RMS resulted in rapid neoplasms regression and normalisation of DIC parameters.

Conclusion Clinical presentation of childhood RMS masquerading as acute leukaemia is unique and poses diagnostic problems, especially in patients with DIC-related haemorrhages. RMS should be included in differential diagnosis of any case presenting as a systemic disease with BM infiltration of cells mimicking leukemic blasts, but lacking lymphoid and myeloid antigens in immunophenotyping by flow cytometry.