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Pseudo-Chediak-Higashi granules in myeloid cells in therapy-related AML with *RUNX1-RUNX1T1*

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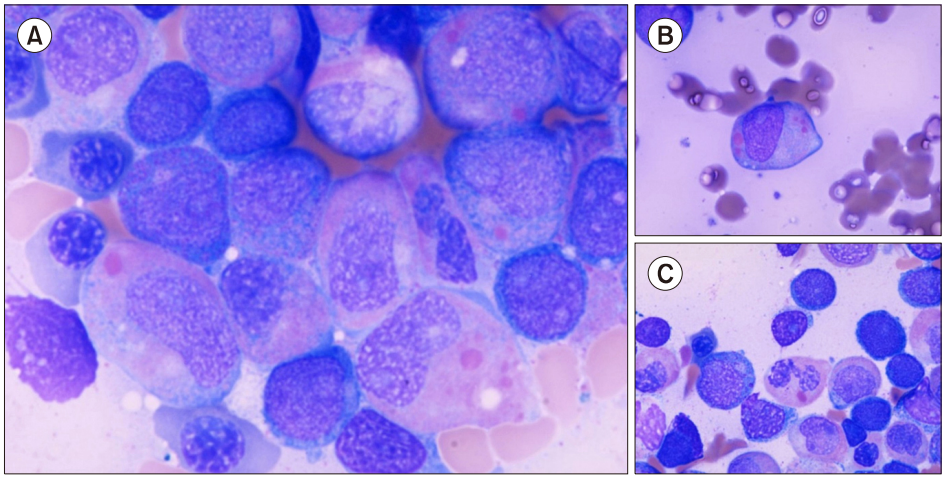

In some acute myeloid leukemia (AML), morphological characteristics and gene rearrangement are closely related. AML with t(8;21)(q22;q22.1);*RUNX1-RUNX1T1* is characterized by perinuclear clearing (hofs), very large granules (pseudo-Chediak-Higashi granules), and a single long and sharp Auer rod with tapered ends in blasts. A 34-year-old woman presented with dyspnea for 2 weeks. She had a history of breast cancer treated with six cycles of chemotherapy and radiotherapy. Her initial white blood cell count was 22.0×10^9/L (55% blasts), hemoglobin level was 4.3 g/dL, and platelet count was 14×10^9/L. Bone marrow was packed with blasts (46% of all nucleated cells) and myeloid cells. Orange-pink granules/globules were present in some blasts and myeloid cells, and some myeloid cells showed homogeneous pink cytoplasm **(A–C)**. Flow cytometry revealed that the blasts were positive for CD13, CD33, CD34, CD56, CD117, HLA-DR, and cytoplasmic myeloperoxidase. Her karyotype was 45,X,-X,del(2)(q33), t(8;21)(q22;q22)[9]/47,XX,+4,t(8;21)(q22;q22)[5]/46,XX[6]. Multiplex reverse transcription-polymerase chain reaction using HemaVision kit revealed the presence of *RUNX1-RUNX1T1* fusion transcripts. The patient was diagnosed with therapy-related AML with *RUNX1-RUNX1T1* based on the 2016 World Health Organization classification. When pseudo-Chediak-Higashi granules are observed in blasts and/or myeloid cells, the presence of *RUNX1-RUNX1T1* rearrangement should be strongly suspected.