A 13 year-old girl with a mass in the left breast and left-sided facial paralysis

CLINICAL HISTORY

A 13 year-old girl was admitted to our department with a mass in her left breast and inability to close her left eye. Her medical history revealed an operation for a lumbar mass in L5 vertebral corpus three years ago when she was diagnosed to have leukemia. Combined chemotherapy and local spinal radiotherapy provided remission for about one and a half years.

In the physical examination, a lobulated mass of 10x5cm in diameters in the left breast and left-sided facial paralysis were observed. In the blood analysis, hemoglobin 9,9 g/dl, leukocyte count 17.200/mm³ with 74% blast cells, platelet count 8.000/mm³ were found. Bone marrow aspirate revealed 100% blast cells, both myeloblasts and monoblasts (Figure 1). Cytochemistry studies showed both peroxidase and non-specific esterase positivity. Blast cells were positive for intracytoplasmic MPO, CD15, CD34 and HLA-DR. Cerebrospinal fluid examination was consistent with meningeal involvement. Computed axial tomography scan of the brain revealed a mass of 3.5 cm in diameter in the left temporal region (Figure 2). Ultrasonography showed a large mass in the left breast with lobulated contours and heterogenous echo texture. Magnetic resonance findings of the breast demonstrated a mass 9x4 cm in diameter in the left breast (Figure 3). The aspiration material, obtained from the breast mass, revealed myeloblasts and monoblasts (Figure 4). Cytogenetic analysis showed 46, XX, t (8;21). (q22, q22).

What is your diagnosis?

PATHOLOGIC DIAGNOSIS

AML associated with extramedullary myeloid tumors (Myeloblastomas or granulocytic sarcomas)
DISCUSSION

An extramedullary myeloid tumor (EMT) is a gross mass of leukemic tissue at a site other than the bone marrow. The incidence of EMT is reported to be between 20% and 50% at the time of diagnosis of childhood AML (1,2). The highest incidence has been reported in the Middle East and Sub-Saharan Africa (3,4). EMT has been reported to occur more frequently in AML in association with the t (8;21) (q22; q22) or inv (16) (p 13; q22) cytogenetic abnormalities, M2, M4 or M5 morphology, CD56 positive cell phenotype and in children than in adults (1,2). Iizuka Y et al. (5) reported that AML patients in whom CD56 was expressed on the leukemic cells showed a greater incidence of development of myeloblastoma and CD56 positive AML patients should be carefully monitored for signs of myeloblastoma formation. EMT has been observed at orbit, maxilla paraspinal region, skin, subcutaneous tissue, gingiva, kidneys, ovary, testis and gastrointestinal tract (2,3,6). We observed extramedullary myeloid tumor at diagnosis as a poor prognostic factor but it should also be noted that the presence of EMT at diagnosis had no significant effect on event-free survival because of intensive treatment regimens (2,3,6,7).

References