Letter to the Editor

ALVEOLAR RHABDOMYOSARCOMA IN A CHILD WITH DIFFUSE BONE MARROW INVOLVEMENT AND CHROMOSOMAL TRANSLOCATION (2;13)

Nur Olgun, MD, Gülersu İrken, MD, Hale Ören, MD, and Bahattin Ören, MD □ Department of Pediatric Hematology and Oncology, Faculty of Medicine, Dokuz Eylül University, İzmir, Turkey

Oğuz Altungöz, MD □ Department of Medical Biology, Faculty of Medicine, Dokuz Eylül University, İzmir, Turkey

Emek Özen, MD □ Department of Pathology, Faculty of Medicine, Dokuz Eylül University, İzmir, Turkey

Meral Sakizli, MD □ Department of Medical Biology, Faculty of Medicine, Dokuz Eylül University, İzmir, Turkey

Namik Çevik, MD □ Department of Pediatric Hematology and Oncology, Faculty of Medicine, Dokuz Eylül University, İzmir, Turkey

Rhabdomyosarcoma (RMS) is a highly malignant soft tissue sarcoma that occurs primarily in childhood, with a high incidence in children under 10 years of age. Approximately 6% to 16% of patients with RMS have bone marrow involvement at presentation.[1,2] There are only a few published descriptions of karyotypes from human RMS; most concern alveolar RMS.[3-7] We report a child diagnosed as alveolar RMS, demonstrating pseudorosette formation and erythrophagocytosis in bone marrow aspiration, with chromosomal aberration (such as translocation [2;13]) plus double minute chromosomes in chromosomal analysis.

CASE REPORT

A 3-year-old white girl was admitted to our hospital with difficulty in closing the right eye, a shift to the left of her lips, and gait disturbance. She had a mass in the right auditory canal and a solitary mass of 4 × 4 cm in diameter at the postero-lateral side of the left foot. She was also found to have

Copyright © 1992 by Hemisphere Publishing Corporation

paralysis of the 6th, 7th, and 12th cranial nerves. Other physical findings were unremarkable.

Laboratory investigations revealed normochrome normocytic anemia with a hemoglobin concentration of 8.5 mg/dL. White blood cell count, neuron specific enolase, urine, and serum vanillylmandelic acid (VMA), and serum ferritin were normal. Chest radiographs and computerized tomography of the thorax demonstrated a 3 × 4 × 2 cm solitary mass lesion in the left hemithorax showing destruction at the posterior side of the third rib. On cranial computerized tomography scan, a tumor mass extending from temporal fossae to the mastoid and jugular fossae, accompanied by the destruction of the petrose bone, was seen. Involvement of tympanic cavity was also noted. Bone survey revealed metastatic lesions at the proximal part of the right humerus and at the diaphysis of the left femur.

In the bone marrow aspiration, 80% of the cells were large, atypical, and immature blasts with large and irregular nuclei, containing several nucleoli and homogeneous granular chromatins. These cells had numerous cytoplasmic vacuoles and they were arranged in small clumps resembling pseudorosettes. Erythrophagocytosis by tumor cells was also present.

Cytogenetic studies performed on the bone marrow aspirate revealed a chromosomal translocation (2;13) in 98% of the metaphases analyzed. Several cells were hypodiploid with chromosomal loss of number 13 also showing derivative 2. In 60% of the metaphases, double minute (DM) chromosomes were observed. All metaphases with DMs were the ones with translocation (2;13), indicating their presence only in neoplastic cells.

On pathological examination, histological features of alveolar RMS were present. The case was diagnosed as stage IV alveolar RMS, and chemotherapy was started with epirubicine, vincristine, actinomycin D, and cyclophosphamide (pulse EVAC). Complete remission was achieved with the chemotherapy and the chromosomal analysis in the bone marrow aspirate during the remission revealed solely normal stemline cells. However, the parents discontinued the therapy and we were informed that the patient died 5 months later.

DISCUSSION

In this case, due to the multiple bone metastases and pseudorosette formation in bone marrow, our initial diagnosis was in favor of neuroblastoma. However, serum and urine VMA and serum neuron specific enolase values are within normal limits. On the other hand, pseudorosette formation and more rarely erythrophagocytosis in the bone marrow as well as multiple bone metastases have also been reported in RMS.[2,8-10] After histopathologic examination of the slides prepared from foot lesion, alveolar RMS was diagnosed. This diagnosis was confirmed by two different pathological centers after mul-

tiple histological reviews. In the literature t(2;13)(q37;q14) have been reported, especially in those cases with alveolar RMS; it was suggested that this may be a specific chromosomal abnormality for alveolar RMS.[3-6] However, Douglass et al[7] recently identified t(2;13) also in undifferentiated RMS and embryonal RMS.

Results of future cytogenetic studies in cases with RMS will help to clarify how specific the translocation (2;13) is for alveolar RMS.

REFERENCES

1. Anner RM, Drewkind B. Frequency and significance of bone marrow involvement by metastatic solid tumors. *Cancer.* 1977;39:1337-1344.
2. Ruymann FB, Newton WA, Ragab AH, et al. Bone marrow metastases at diagnosis in children and adolescents with rhabdomyosarcoma: a report from the Intergroup Rhabdomyosarcoma Study. *Cancer.* 1984;53:368-373.
3. Brichard B, Gosseye S, Verellen-Dumoulin C, et al. A disseminated alveolar rhabdomyosarcoma in a 9-year-old boy disclosed by chromosomal translocation (2;13) (q35;q14). *Pediatr Hematol Oncol.* 1991;8:215-219.
4. Lizard-Nacol S, Mugneret F, Volk C, et al. Translocation (2;13) (q37;q14) in alveolar rhabdomyosarcoma: a new case. *Cancer Genet Cytogenet.* 1987;25:373-374.
5. Seidal T, Mark J, Hagmar B, et al. Alveolar rhabdomyosarcoma: a cytogenetic and correlated cytological and histological study. *Acta Pathol Microbiol Immunol Scand [A].* 1982;90:345-354.
6. Turc-Carel C, Lizard-Nacal S, Justraba E, et al. Consistent chromosomal translocation in alveolar rhabdomyosarcoma. *Cancer Genet Cytogenet.* 1986;19:361-362.
7. Douglass EC, Valentine M, Etcubanas E, et al. A specific chromosomal abnormality in rhabdomyosarcoma. *Cytogenet Cell Genet.* 1987;45:148-155.
8. Etcubanas E, Peiper S, Stass S, et al. Rhabdomyosarcoma, presenting as disseminated malignancy from an unknown primary site: a retrospective study of ten pediatric cases. *Medical Pediatr Oncol.* 1989;17:39-44.
9. Huntrakoon M, Callaway LA, Vergara GG. Systemic rhabdomyosarcoma presenting as leukemia: case report with ultrastructural study and reviews. *J Surg Oncol.* 1987;35:259-265.
10. de la Serna FJ, Martines MA, Valdes MD, et al. Rhabdomyosarcoma presenting with diffuse bone marrow involvement, hypercalcemia and renal failure. *Medical Pediatr Oncol.* 1988;16:123-127.

Received January 2, 1992
Accepted March 11, 1992

Address correspondence to Nur Olgun, MD, Mithatpaşa Caddesi, Mehveş Apt. No: 1156/7, Güzelyali, İzmir, Turkey.