Conflict of Interest: The author of this paper has no conflicts of interest, including specific financial interests, relationships, and/or affiliations relevant to the subject matter or materials included.

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Isolated Mediastinal Myeloid Sarcoma after NPM1-Positive Pediatric Acute Myeloid Leukemia

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To the Editor,

Myeloid sarcoma (MS) is a rare extramedullary mass that consists of immature myeloid cells. The most common locations are the soft tissue, bone, periosteum, orbit, and lymph nodes [1,2]. Mediastinal involvement is very rare and most commonly reported with concurrent bone marrow involvement [3]. Herein we present a previously treated nucleophosmin (NPM1)-positive acute myeloid leukemia (AML) patient who later presented with isolated mediastinal MS.

A 9-year-old female patient presented with fatigue and weakness. Physical examination revealed no pathological findings. Blood tests demonstrated hemoglobin of 12.2 g/dL, hyperleukocytosis (100,500/µL), and thrombocytopenia (43,000/µL) with 88% blasts in the peripheral blood smear. Bone marrow aspirate revealed 90% blasts with M1 subtype. Treatment was started according to the AML-BFM 2012 protocol. Conventional cytogenetic analysis failed due to lack of spontaneous mitosis and fluorescent in situ (FISH) analysis for t(8;21), inv(16), t(15;17), and t(9,22) from bone marrow samples revealed negative results. Molecular genetic analysis in the peripheral blood showed NPM1 positivity and FLT3-ITD negativity. Conventional cytogenetic analysis from the bone marrow was within normal limits, while NPM1 could not be studied from bone marrow. Her previous CT scans that were performed for investigation of invasive pulmonary aspergillosis were all normal. Fine-needle aspiration biopsy of the mass was performed; histopathological examination revealed myeloblasts that were positive for myeloperoxidase, CD15, and CD33. Microscopic examination of the imprint of the biopsy also revealed myeloblasts of M1 subtype (Wright stain). Major reduction in tumor mass (7 mm residual tumor) and NPM1 negativity were achieved after one block of FLAG (fludarabine, cytarabine, filgrastim) and two blocks of FLAG-mitoxantrone. The patient underwent successful bone marrow transplantation from a matched unrelated donor and has been in remission for one year.

MS of the mediastinum is very rare; most of the cases have been reported as initial presentation with concurrent bone marrow involvement [3,4,5]. MS as a relapse has been more frequently reported in post-transplant patients compared to those treated without allogeneic hematopoietic stem cell transplantation [6,7]. Our patient is unique as she presented with isolated mediastinal MS after chemotherapy treatment. Another important point about our patient is that the NPM1 positivity was detected at...
the same time as MS. The incidence of MS has been known to be higher in certain cytogenetic abnormalities, in particular t(8;21) [1,6]. Falini et al. [8], in their study with 181 MS samples, identified NPM1 mutations as the most frequent molecular lesion in MS, defining the molecular status in 15% of cases. Our patient was negative for t(8:21) but had NPM1 positivity.

In conclusion, even though NPM1 is not a poor prognostic factor for AML, it should be kept in mind that patients with NPM1 positivity may later present with MS, as in the case of our patient, who presented with isolated MS of the mediastinum months after cessation of chemotherapy.

Keywords: Acute myeloid leukemia, Myeloid sarcoma, Mediastinal mass, NPM1

Anahtar Sözcükler: Akut myeloid lösemi, Myeloid sarkom, Mediastinal kitle, NPM1

Informed Consent: Written informed consent for publication was obtained from the patient and her parents.

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