Amyloidosis and Myelodysplastic Syndrome

Satoko Oka¹, Kazuo Muroi¹, Masaki Toshima¹, Tadashi Nagai¹, Nobuyuki Kanai², Tatsuo Morita³ and Keiya Ozawa¹

Key words: myelodysplastic syndrome, amyloidosis

(DOI: 10.2169/internalmedicine.47.0533)

A 61-year-old woman presented with painless gross hematuria in the Department of Urology, Jichi Medical University Hospital in January 1999. She had no significant past medical history. There were no hematological disorders including amyloidosis in her family. Physical examination did not show any abnormalities including skin and heart.

¹Division of Hematology, Department of Medicine, Jichi Medical University, Shimotsuke, ²Department of Pathology, Jichi Medical University, Shimotsuke and ³Department of Urology, Jichi Medical University, Shimotsuke

Received for publication August 16, 2007; Accepted for publication October 9, 2007
Correspondence to Dr. Kazuo Muroi, muroi-kz@jichi.ac.jp
Cystoscopy demonstrated nodular masses with hemorrhagic mucosal surfaces in the urinary bladder. Pathological examination of the biopsy specimens showed the presence of amyloid deposits stained with Congo red (Picture 1a). The potassium permanganate treatment did not block Congo red staining of the deposits (Picture 1b). The amyloid deposits were not stained with anti-human serum amyloid A monoclonal antibody. Amyloid was not found in the upper gastrointestinal tract, while a mild form of it was detected in the rectal mucosa. Treatment of amyloidosis with bis-coclaurine alkaloid cepharanthine was started. She was consulted for amyloidosis by a hematologist in the Division of Hematology in April 2001. Laboratory studies showed a hemoglobin level of 13.3 g/dl, a platelet count of 14.2×10^4/μl, and a white blood cell count of 2.7×10^3/μl with normal differential. Monoclonal protein was not shown by serum protein electrophoresis analysis. Bone marrow aspiration demonstrated mild trilineage morphological abnormalities without increase in blasts and plasma cells. Chromosomal analysis of the specimens showed 46, XX, -20, +mar (7 cells/8 cells). Ultrasound sonography did not reveal any abnormalities in the abdomen, including the liver, spleen, kidneys and lymph nodes. Electrocardiography did not show any abnormalities. She was diagnosed as having myelodysplastic syndrome (MDS). Five years after the diagnosis, bone marrow aspiration was performed to determine disease status of MDS; mild abnormalities in cell maturation of granulocytes, erythroblasts and megakaryocytes without an increase in blasts persisted (Picture 2), and karyotypic abnormalities were the same as those at the initial presentation. Peripheral blood cell counts are almost within normal ranges except for a slight decrease in platelets. Cepharanthine has been administered for eight years and macrohematuria did not occur in the previous year. New lesions associated with amyloidosis were not observed.

AL amyloidosis is one of the most common amyloidoses; it can be found in a localized and a systemic form. Primary systemic amyloidosis is usually associated with the deposition of immunoglobulin light chains, with or without an underlying plasma-cell dyscrasia. The localized form of AL amyloidosis in the urinary bladder has been reported (1). A few cases of amyloidosis associated with MDS have been reported: one case was MDS with localized amyloidosis of the colon, however the amyloid types were not determined in this case (2). Two cases were MDS with systemic AA amyloidosis (3). Therefore, this is the first case of MDS complicated by non-AA amyloidosis. The pathogenesis for the development of amyloidosis and MDS in the present patient remains unclear. A common abnormal hemopoietic stem cell might cause both AL amyloidosis and MDS. Alternatively, since MDS is thought to arise from mutations in primitive hemopoietic stem cells, abnormal antigen presentation, abnormal T- and B-cell interaction, or abnormal macrophage function may cause immune dysregulation which leads to the development of amyloidosis. To identify the relationship between amyloidosis and MDS, it is necessary to collect cases of the two concomitant disorders.

References


© 2008 The Japanese Society of Internal Medicine
http://www.naika.or.jp/imindex.html