Multiple Myeloma Associated with Amyloidosis and t(1;20)(q21;q11) Translocation

KOROKU OTOKIDA, HIROAKI YOSHIDA, YOSHIMI MIZUNUMA,
MICHIO YAMADA and KATSUHIKO HIRAMORI

*The Second Department of Internal Medicine, Iwate
Medical University School of Medicine, Morioka 020*

OTOKIDA, K., YOSHIDA, H., MIZUNUMA, Y., YAMADA, M. and HIRAMORI, K. *Multiple Myeloma Associated with Amyloidosis and t(1;20)(q21;q11) Translocation.* Tohoku J. Exp. Med., 1990, 162 (4), 363-365 — A 59-year-old woman with nephrotic syndrome was diagnosed as having primary amyloidosis based on the detection of amyloid deposition (AL-protein) in the esophagus and kidneys. Bone marrow aspirate showed plasmocytic proliferation, leading to a diagnosis of multiple myeloma (IgG λ -type). In addition, a very rare translocation t(1;20)(q21;q11) was seen by chromosomal analysis of both the bone marrow and peripheral blood. — multiple myeloma; amyloidosis; chromosomal abnormality

Only a few cases of t(1;20) translocation have been reported: A patient with chronic myelocytic leukemia with Philadelphia chromosome (Ph¹) was found to have an additional chromosomal abnormality t(1;20)(q21;q13) in 1978 (Norman and Boucher). Then, an azoospermic man with t(1;20)(q21;q13) was reported by Antonelli et al. in 1983. Madan and Kleinhout (1987) subsequently showed that the women of a large family that suffered from first trimester abortions had t(1;20)(p36;p11). Most recently, Weh et al. (1990) have reported two patients with multiple myeloma and t(1;20)(q12.3;p13). We here report a woman with multiple myeloma, the nephrotic syndrome and systemic amyloidosis, who also had the chromosomal abnormalities t(1;20)(q21;q11).

CASE REPORT

A 59-year-old house-wife was admitted for treatment of the nephrotic syndrome on 26 May, 1989. The hemogram was normal and the Westergren sedimentation rate was increased. Liver and kidney function was normal. Urinary protein excretion ranged between 10 and 15 g/day, with Bence-Jones protein being detected by immuno-electrophoresis. Her serum total protein level was low (4.9 g/100 ml), but there was an increase of γ -globulin (16.9%). This M-protein was shown to be an IgG λ -type paraprotein by immuno-electrophoresis. The serum IgG level was elevated (1,473 mg/100 ml), while both the IgA (47 mg/100 ml) and IgM (61 mg/100 ml) levels were low. Bone marrow aspiration microscopically showed the proliferation of plasmocytes (18.0%) with scattered plasmocytic packets, leading to a diagnosis of multiple myeloma. Needle biopsy of the kidney indicated amyloidosis; with Congo Red staining AL-type amyloid deposition along the capillary walls thickened the basement membrane, resulting in the nephrotic syndrome. Chromosomal analysis by G-banding was performed after the culture of the bone marrow

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Fig. 1. G-banding chromosomal analysis of the peripheral blood showed karyotype of 46, XX, t(1;20)(q21;q11) in the patient.

without phytohemagglutinin (PHA) and peripheral blood with PHA, because no analysable mitosis of the peripheral blood had happened in the first culture without PHA. The karyotype of all the analyzed cells was 46, XX, t(1;20)(q21;q11), which was identical in both the bone marrow and peripheral blood, as shown in the Fig. 1.

The patient's hypoalbuminemia was improved by bed rest and an increased food intake, and she was discharged in July 1989. However, serum protein level decreased again and she was readmitted in December 1989. Marked generalized edema and impairment of renal function were noted, and she was maintained on hemodialysis. Amyloid deposition was seen in the veriform appendix which was removed at an operation for appendicitis, as well as in biopsies of the skin and esophagus. No osteolytic changes in the bones were observed, despite her not being given chemotherapy for multiple myeloma.

DISCUSSION

Multiple myeloma, chronic myelocytic leukemia (CML) and acute non-lymphocytic leukemia are frequently associated with chromosomal aberrations (Dewald et al. 1985; Rowley and Golomb 1984). However, the presence of translocation t(1;20) in patients with leukemia or multiple myeloma was not documented, except for one patient with Ph⁺-positive CML and an additional t(1;20) (Norman and Boucher 1978). Weh et al. (1990) reported the first two patients with multiple myeloma and t(1;20).

It is of interest that Le Beau et al. (1984) found two oncogene (*c-src*) loci on the human chromosomes 1 and 20, at 1p34-p36 and 20q12-q13. However, these sites were completely different from the translocated breakpoints in the patient with CML (1q21 and 20q13) reported by Norman and Boucher (1978), the two patients with multiple myeloma (1q12.3 and 20p13) reported by Weh et al. (1990) and in our patient with multiple myeloma (1q21 and 20q11) described here. It is likely that the incidence of t(1;20) translocation is higher in patients with multiple myeloma than in leukemic patients, but further chromosomal analysis studies are necessary to demonstrate whether this is so.

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