Deletion of the long arm of chromosome 20 (del 20q) has been observed in patients with myelodysplastic syndrome (MDS) or myeloid malignancies. We experienced an MDS female case of del 20q accompanied by clusters of plasmacytic cells in bone marrow. Her bone marrow cells showed morphological abnormalities in three lineages and the chromosomal abnormality of 46, XX, del (20) (q11.2q13.3). Although the percentage of plasma cells was low in free cells, such cells showed nuclear abnormalities. In bone marrow clots, we also observed clusters of anti-CD38 and anti-CD138 antibody-positive cells. According to the FAB or WHO classification, the diagnosis was unclear. Therefore, we were obliged to term this case as MDS with plasma cell dysplasia. This patient was considered to be a rare case of MDS related to abnormalities in myeloid and B-lymphoid cells. [J Clin Exp Hematopathol 51(2): 141-145, 2011]

**Keywords:** myelodysplastic syndrome, del (20q), plasma cell, CD38, CD138

**INTRODUCTION**

Myelodysplastic syndrome (MDS) is a hematological disease derived from abnormal maturation and differentiation of stem cells and is clinically manifested as cytopenia. MDS, especially a case with deletion of the long arm of chromosome 20 (del 20q), has been generally thought to be a result of abnormal maturation in the myeloid lineage. We report herein a rare MDS case showing del 20q with hypo-globulinemia, no M-protein in serum, no Bence-Jones proteinuria, and no high percentage of plasma cells among bone marrow free cells, but with plasma cell dysplasia in bone marrow clots. The relatively low percentage of plasma cells among bone marrow free cells seemed to mask the B-cell abnormality because clusters of plasmacytic cells positive for anti-CD38 and anti-CD138 antibody were observed in bone marrow clots. We had to draw our own conclusions about diagnosis in this case, namely, MDS of del 20q with plasma cell dysplasia that showed abnormalities of both myeloid and B-lymphoid lineages. No examination data were obtained to show that this case was a B-cell malignant disorder.
$189 \text{ g/dL}$, and $479 \text{ ng/mL}$, respectively, in which only ferritin was elevated. Blood urea nitrogen and creatinine were $38.1 \text{ mg/dL}$ and $2.37 \text{ mg/dL}$, respectively. Her serum albumin was low ($3.0 \text{ g/dL}$; normal range, $>3.7 \text{ g/dL}$), indicating malnutrition. Serum immunoglobulins (Ig) consisting of IgG at a concentration of $740 \text{ mg/dL}$ but with a low level of IgG4 (IgG1, 61.4% ; IgG2, 34.33% ; IgG3, 3.42% ; IgG4, < 0.31%), IgA at $146 \text{ mg/dL}$, IgM at $185 \text{ mg/dL}$, IgE at 121 IU/mL, and IgD at 1.7 mg/dL (the normal ranges of the 5 classes of aforementioned serum Ig are 870~1,700, 110~470, 23~250, 0~170, and < 9, respectively) were observed. Sedimentation test (ESR), C-reactive protein, anti-nuclear antibody, and anti-DNA antibody were > 140 mm/hr, 4.8 mg/dL, × 80, and × 160, respectively. D-dimer and thrombin/anti-thrombin III complex were elevated (3.6 μg/dL and 11.6 ng/mL) because of hyper-coagulation induced by bone fracture. Since her fracture occurred in an unusual region and by an unexpected accident, we performed bone marrow aspiration given her anemia, suspecting a myeloma-like disorder. Our orthopedist did not identify any abnormal lesion in her bone X-ray pictures. Free cells in aspirated bone marrow revealed hypocellularity with only 1.2% of plasma cells showing normal lobulation or pocket in free cells. Such plasma cells could not be light-microscopically qualified as typical myeloma cells. Morphological abnormalities of blood cells in three lineages were also observed. Reticulum cell possessed iron in the cytoplasm (Fig. 1). A chromosomal abnormality was shown as 46, XX, del (20) (q11.2;q13.3) (18 cells/20 analyzed cells, Fig. 2). Moreover, in the aspirated bone marrow clots, the clusters of plasmacytic cells were found to be positive for anti-CD138 or anti-CD38 antibody (Fig. 3). Urinary Bence-Jones protein was negative. Electrophoresis of serum did not reveal any evidence of M-protein.

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**Fig. 1.** Bone marrow smear (May-Grünwald-Giemsa stain). (**1a**) An indistinct cell. (**1b**) Micromegakaryocyte-like cell. (**1c**) Nuclear segmented erythroblast. (**1d**) Nuclear hypersegmented neutrophil with few granules. (**1e**) Reticulum cell (arrow indicates iron-phagocytizing cytoplasm). (**1f**) Lymphoplasmacytic cell with nuclear pocket (arrow). (**1g**) Neutrophil with pseudo-Pelger-Huët nucleus. (**1h**) Plasma cell with lobulate nucleus.
DISCUSSION

This case showed morphological abnormalities of blood cells in three lineages with abnormal iron metabolism indicated by elevated ferritin in serum and by iron-positive staining in the cytoplasm of reticulum cells. Although the patient showed normocytic anemia, slightly renal dysfunction, and malnutrition, the iron-containing reticulum cells on her bone marrow smears showed that her anemia was refractory against iron supplementation. Her chromosomal abnormality, del (20) (q11.2q13.3), is reported to occur predominantly in the myeloid lineage. Therefore, she was initially diagnosed as having refractory anemia or cytopenia in MDS according to the FAB or WHO classification. Although the increase of plasma cells could not be counted in her bone marrow free cells, plasma cells showed several nuclear aberrations. Her bone fracture at her pubic arch, which was attributed to the slight vibration of a car seat that occurred during a ride in a car being driven on an asphalt-paved road, suggested that she had deteriorated to the point at which her bone was pathologically vulnerable to fracture. Clusters of plasmacytic cells positive for anti-CD38 or anti-CD138 antibody were observed in her bone marrow clots. Such plasmacytic cells might be considered to belong to a certain category of myeloma cells, as supported by the observation that rabbit cells were transformed into CD138-expressing malignant cells when primary human myeloma cells were injected into an immunodeficient rabbit. In this case, we could not identify the plasmacytic cells as typical myeloma ones in multiple myeloma. Meanwhile, the chromosomal abnormality of del 20q has been considered to be a common MDS-associated cytogenic change concerning the myeloid lineage. This chromosomal change was also presented in myeloproliferative disorders (MPD) such as polycythemia vera and essential thrombocythemia, which are not disorders in the myeloid lineage. In the lymphoid lineage, some del 20q-MDS cases with myeloma, with monoclonal gammopathy of undetermined significance, and with small cell lymphocytic lymphoma were also reported. Recently, an uncommon case of chronic myeloid leukemia with myeloma was reported in an elderly woman. However, almost all of these reported MDS cases were complicated with B-cell disorders that had arisen after therapy with alkylating agents. On the other hand, in vitro, both anti-CD38- and del 20q-positive cells were observed in the Epstein-Barr virus-transformed lymphoid cell line from the cells of a del 20q-MDS case. These reports might indicate that the del 20q-associated cytogenic finding is associated with a chromosomal abnormality related to not only myeloid but also other lineages. Alternatively, MDS with B-cell abnormality might be related to a viral infection such as...
Epstein-Barr virus. We cannot explain the significance of the low level of IgG4 in this patient’s serum IgG because this elderly MDS case with both plasmacytic cell dysplasia and a low level of IgG4 is the first of its kind reported globally. The slightly elevated anti-nuclear antibody, anti-DNA antibody, ESR, and C-reactive protein in her serum might be telltale signs of abnormal functions of her B-cells, although the localized inflammation upon her bone fracture would have induced several immunological reactions in her body. The clusters of anti-CD38- and anti-CD138-positive cells exist not as free cells but in clots accompanied by MDS. In general, some elderly subjects with or without MDS show immunological abnormalities as previously discussed. We could not draw any conclusions on the causal relationship between her immunological abnormalities and anti-CD138- or anti-CD38-positive cells.

When we consider this rare MDS case, there is some possibility that a certain type of MDS might be related to B-cell abnormality as a pathological factor. Nonetheless, we experienced a truly rare case that showed coincidental abnormal transformations in myeloid precursor cells and B-cells.

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