

INTRODUCTION

Deletion of the long arm in chromosome 5 in myelodysplastic syndrome (MDS) ranges in frequency from 16 to 28%. A distinct syndrome is recognized by the WHO in which this deletion is the sole abnormality. Myelodysplastic syndrome (MDS) associated with isolated del (5q) is associated with a much better prognosis than other subtypes.

We present a case of a 71 year-old female with persistent macrocytic anemia. A bone marrow was performed which showed morphological evidence of myelodysplasia. Cytogenetics showed deletion of 5q and t(4;22). A diagnosis of refractory cytopenia with multilineage dysplasia was rendered in this case. However, morphological findings and clinical course in this patient are more consistent with MDS with isolated del (5q).

MATERIALS

Peripheral blood and bone marrow specimens were received in pathology. Wright and Hematoxylin-eosin stains were evaluated. Cytogenetics were performed.

RESULTS

Peripheral blood and bone marrow showed features of MDS with isolated del (5q): macrocytic anemia, normal platelet count, hypercellular bone marrow with erythroid dysplasia, marked increase in megakaryocytes with small size and hypolobated nuclei, blast count of 4%, and no ring-sideroblasts (Figs. 2, 3, 4).

Chromosome analysis of 20 cells from bone marrow (Fig. 1) showed 18 cells with interstitial deletion of 5q as the only abnormality, and 2 cells with both del(5q) and t(4;22). According to the WHO criteria, this case is best classified as refractory cytopenia with multilineage dysplasia. Follow-up of this patient 15 months after diagnosis showed unchanged blood counts. The patient has been in stable condition and requires no treatment.

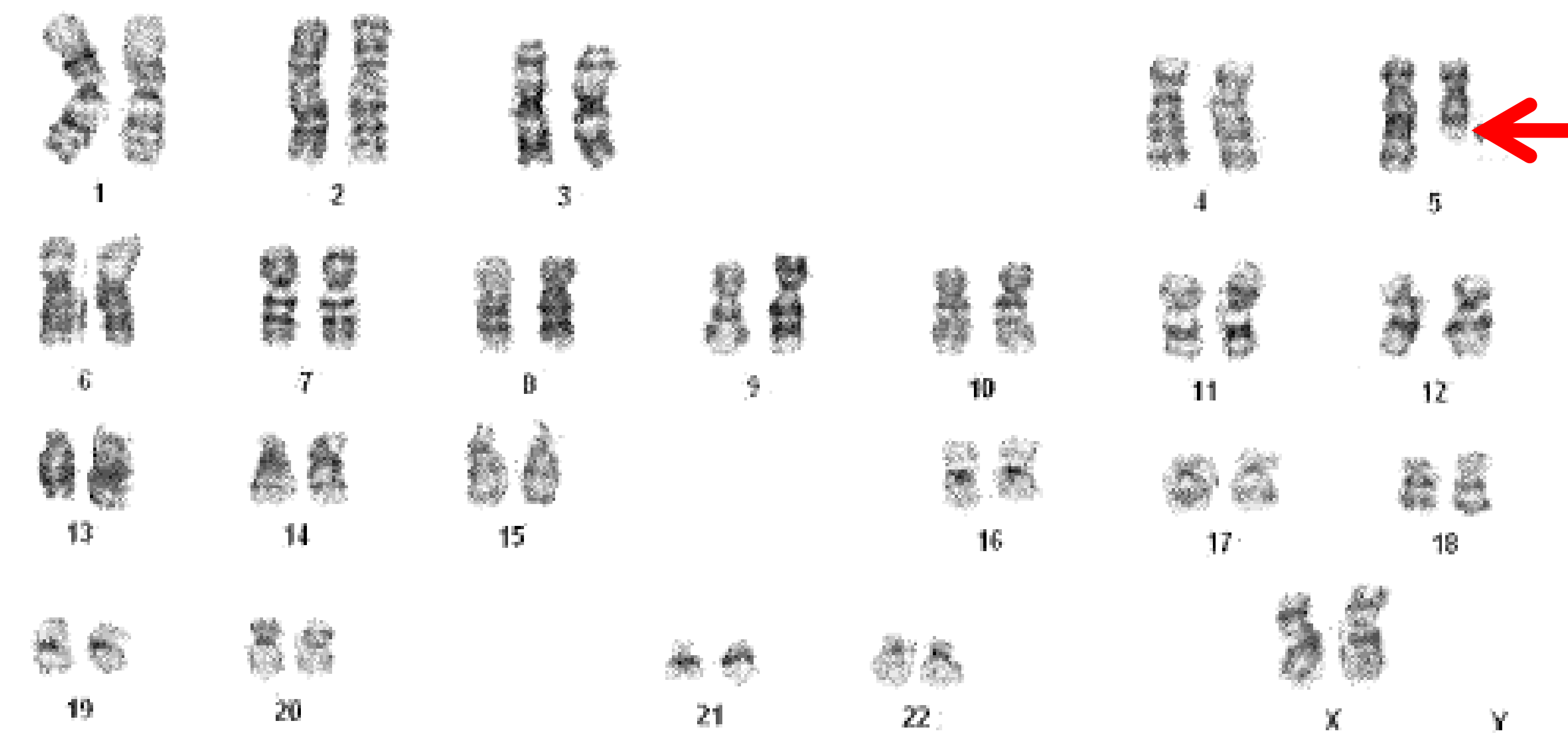


Fig 1. Chromosome analysis

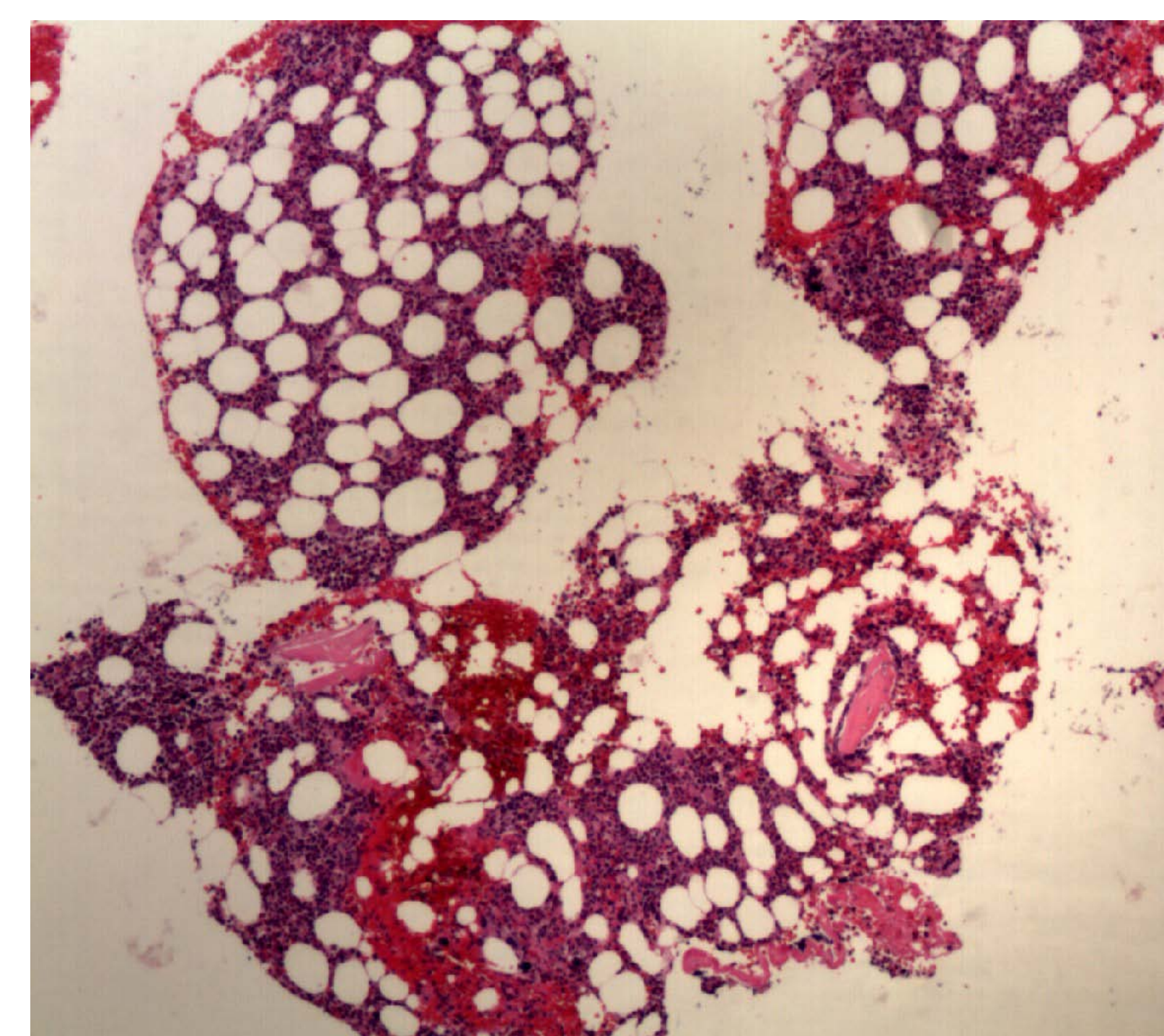


Fig 2. Hypercellular bone marrow biopsy, 10x

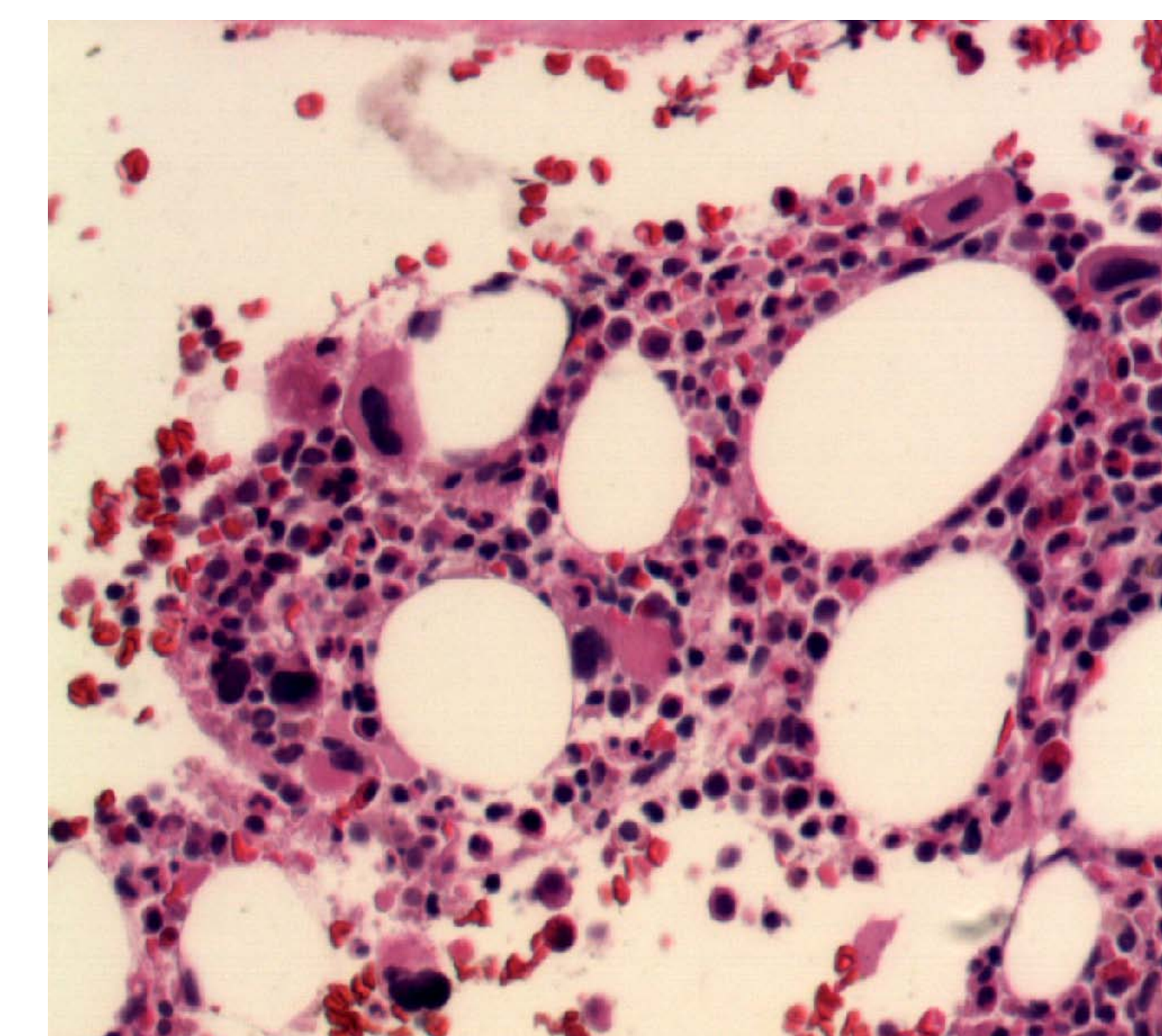


Fig 2. Hypolobated megakaryocytes in biopsy, 20x

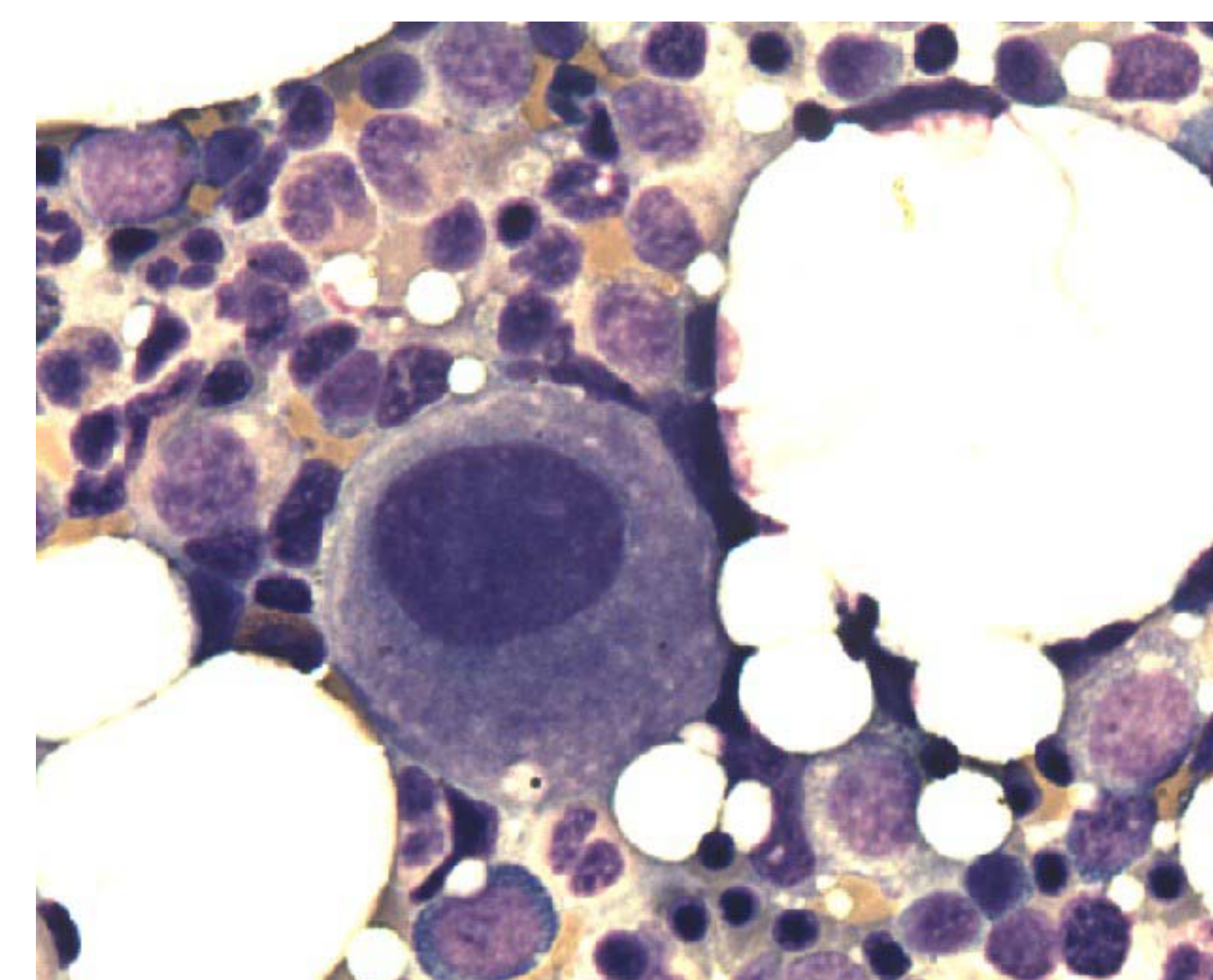


Fig 3. Hypolobated Megakaryocyte in aspirate, 50x

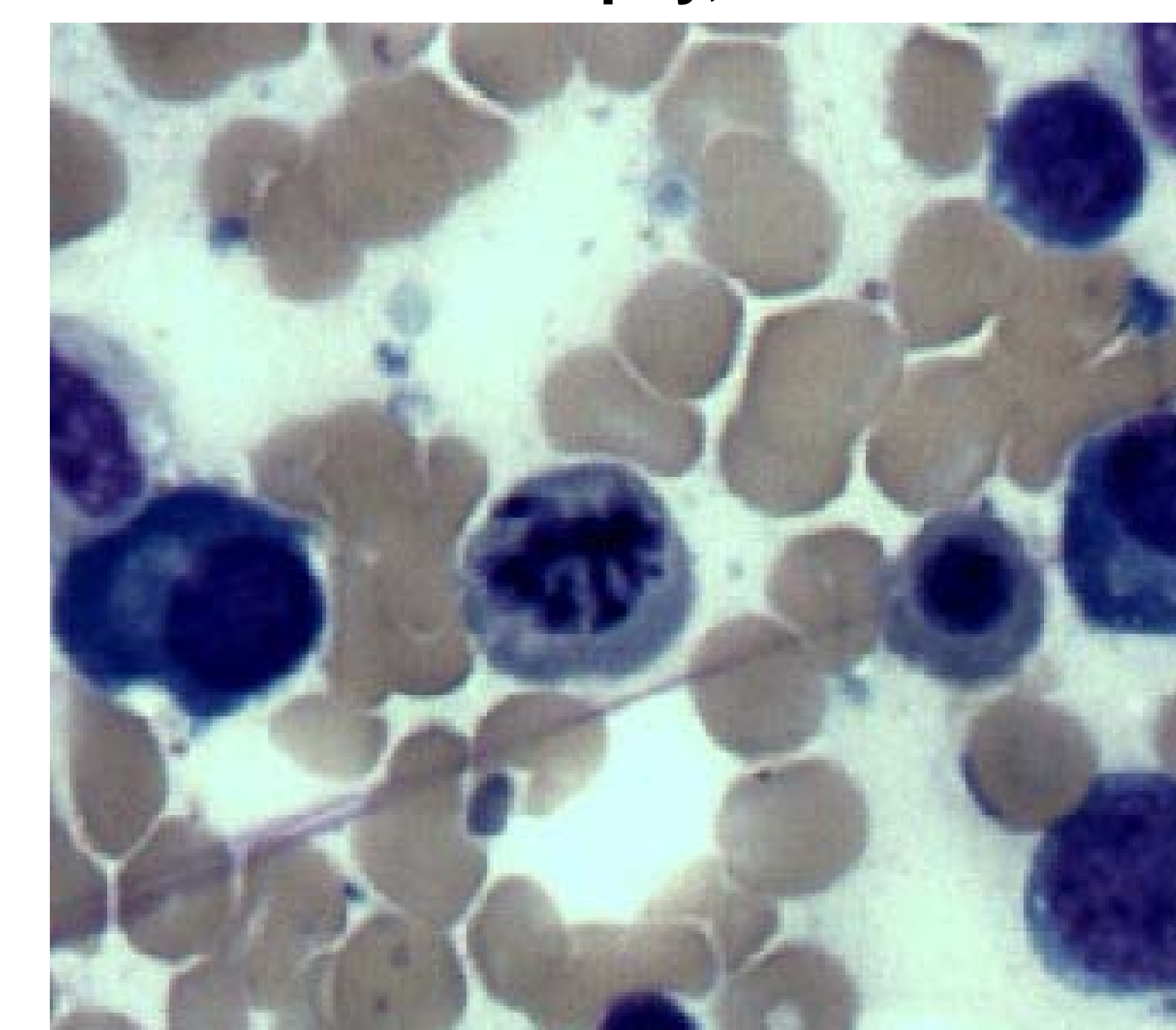


Fig 4. Dysplastic RBC in aspirate, 100x

DISCUSSION

Chromosome analysis of 20 cells from bone marrow (Fig. 1) showed 18 cells with interstitial deletion of 5q as the only abnormality, and 2 cells with both del(5q) and t(4;22). Based on the WHO criteria, a diagnosis of refractory cytopenia with multilineage dysplasia is rendered in this case. However, this patient is only anemic with normal leukocyte count and platelet count. She has been followed up without medical treatment, and her counts have been unchanged for 15 months. The clinical course is therefore more similar to that of MDS with isolated del (5q).

MDS with isolated del 5q usually has macrocytic anemia and adequate or increased platelet count. The bone marrow's characteristic feature are dysplastic erythroids, hypolobated megakaryocytes. Blast count by definition is less than 5%.

Patients with this entity have a long survival. Morbidity is related to the anemia, which can become transfusion dependent. Since these patients usually have increased erythropoietin, treatment with the recombinant form does not work. Treatment with lenalidomide, a thalidomide analog, has been proven successful in many cases, and cytogenetic remission has also been observed.

Progression to acute myeloid leukemia is rare. The addition of cytogenetic abnormalities increases the likelihood of acute leukemia progression and is believed to shorten survival.

REFERENCES

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