A complex karyotype in a case with myelodysplastic syndrome

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Myelodysplastic syndromes (MDS) are a heterogeneous group of clonal hematologic malignancies that are characterized by dysplasia and ineffective hematopoiesis in the myeloid cell lineages, leading to blood cytopenias and a high risk of progression to acute myeloid leukemia. Chromosomal anomalies are observed in 30–50% of de novo MDS patients, like complete or partial loss of chromosome 5 or 7, loss of Y, trisomy 8 and deletions of 11q, 17p or 20q. Patients with complex karyotype or loss of chromosome 7 have poor prognosis; patients with a 5q or 20q deletion as a sole abnormality have good prognosis. The authors present a case of a 69-year-old woman with MDS. The hemogram analysis showed trilinear dysplasia with peripheral cytopenia, anemia and thrombocytopenia and did not present excess of blasts in the bone marrow. Bone marrow cell cultures and GTL banding were performed according to the protocols in the laboratory. FISH were performed to clarify the anomalies detected. Cytogenetic analysis followed the standard guidelines.

The karyotype was: 46,XX,−4, der(11)dup(11?), del(7)(q32), del(12)(p11),+mar [5]/47,X, der(X)t (X;7), del(12)(p11),+mar [2]/46, XX, del(7)(q32), del(12)(p11) [10]/46, XX [4]. This is a complex karyotype with three abnormal cell lines, with clonal evolution. It has two structural abnormalities common to MDS (deletion 7q and 12p) both associated with poor prognosis. The other rearrangements detected are rare and not
associated with MDS. The authors compare the results with those described in the literature.