

Clinical Image

Myelodysplastic Syndrome with Isolated del (5q) Evolved to Acute Myeloid Leukemia LMA-M6 Subtype

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Myelodysplastic syndrome; 5q- syndrome; Acute myeloid leukemia M6

The 5q- syndrome is characterized by a macrocytic anemia with a normal- elevated platelet count, erythroid hypoplasia, large mononuclear (LMN) megakaryocytes and the 5q31 deletion as the sole karyotypic abnormality. It is associated with long survival and

low rate of acute leukemic transformation. A 79-year-old woman presented with blood counts of Hb 98 g/L, VCM 109 fL, neutrophils $1.5 \times 10^9/L$ and platelets $87 \times 10^9/L$. A bone marrow (BM) aspirate showed LMN megakaryocytes (Figure 1a) and a karyotype with the del (5)(q14q33). It was diagnosed of a myelodysplastic syndrome (MDS) with isolated del (5q). Months later, blood counts dropped and a BM study showed 37% of blasts with round nucleus with basophilic and vacuolated cytoplasm (Figure 1 b-d), PAS staining positive (Figure 1 e-f) and immunophenotype with CD34-, CD117+, CD71+, CD105+ (Figure 2a). Karyotype showed clonal evolution (Figure 2b). This is a rare case because of the exceptional transformation of an MDS with isolated del(5) to an acute erythroid leukemia.

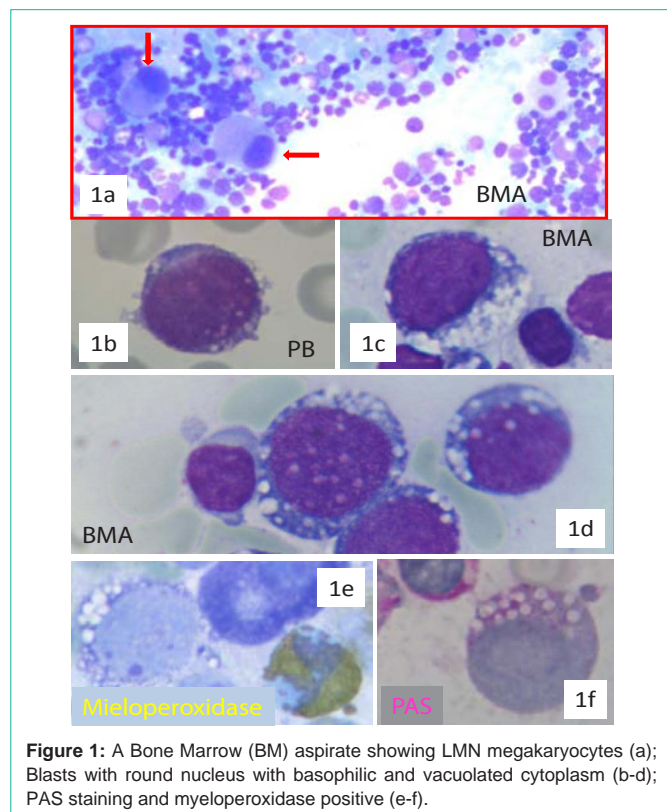


Figure 1: A Bone Marrow (BM) aspirate showing LMN megakaryocytes (a); Blasts with round nucleus with basophilic and vacuolated cytoplasm (b-d); PAS staining and myeloperoxidase positive (e-f).

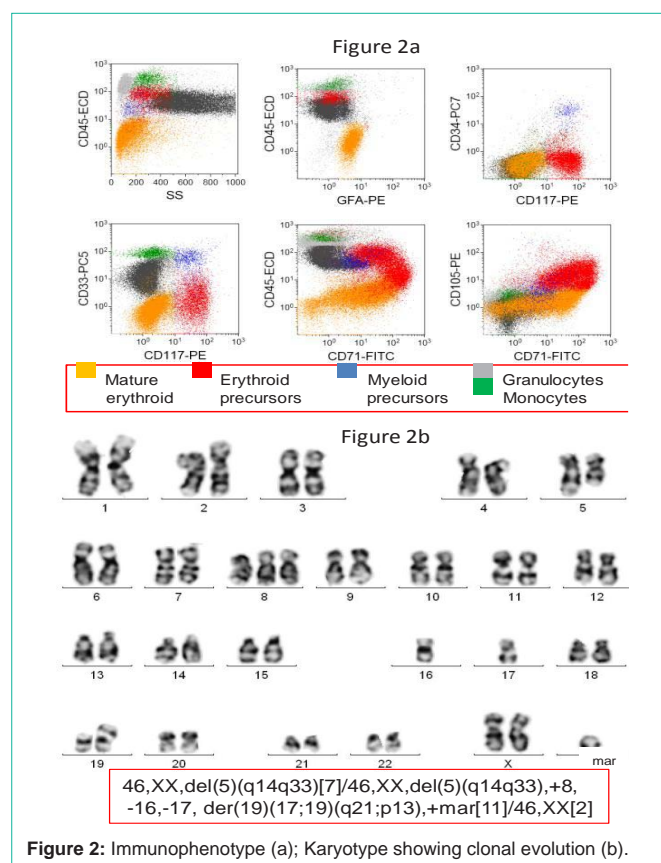


Figure 2: Immunophenotype (a); Karyotype showing clonal evolution (b).