IMAGENS EM HEMATOLOGIA/IMAGES IN HEMATOLOGY

Clinical features and chromosomal abnormality in Myelodysplastic Syndrome of the Refractory Anaemia Subtype

Aspectos clínicos e anormalidade cromossômica em Síndrome Mielodisplásica do Subtipo Anemia Refratária

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Cytogenetic evaluation in myelodysplastic syndrome (MDS) has been considered important not only for clonality determination at diagnosis, but also for prognosis.[1] Recently, the International Prognostic Scoring

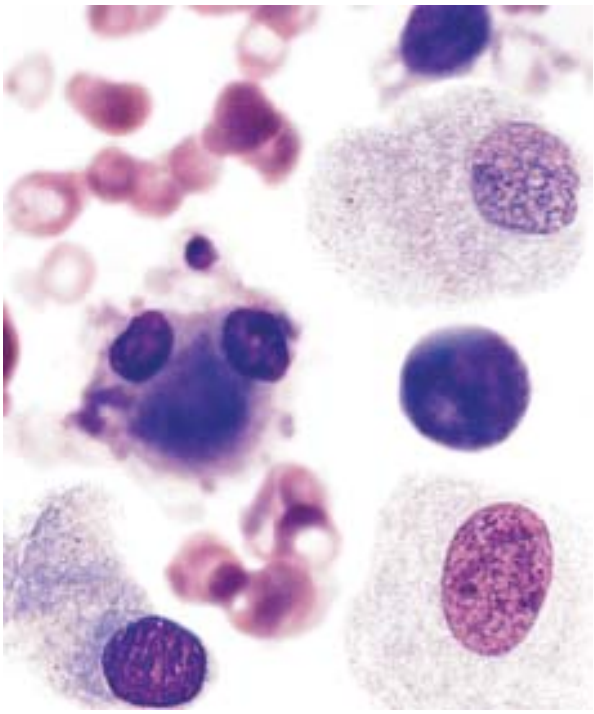

***Fig. 1 –** Small megakaryocytes with hypolobulated nucleus obtained from a bone marrow sample of the myelodysplastic syndrome patient (Romanovsky, 1200x)*

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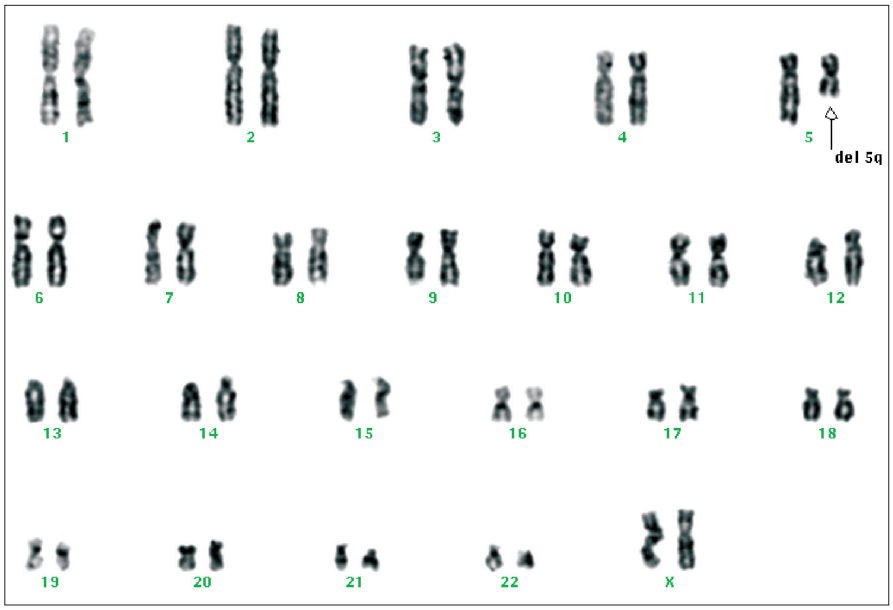

***Fig. 2** – G-banded karyotype obtained from the myelodysplastic syndrome patient at diagnosis: 46, XX, del(5)(q13;q33). The arrow shows the chromosomal abnormality*

System has proposed identified chromosomal abnormalities as one of the most important variables to determine survival and to predict the risk of transformation into acute leukaemia[2]. Loss of material from chromosome 5 as a result of monosomy or deletion is quite common and occurs in 10-30% of patients with MDS who show chromosomal aberrations.[3]

The 5q- syndrome is a MDS with the 5q deletion as the sole karyotypic abnormality and is characterised by refractory macrocytic anaemia, hypolobulated megakaryocytes in bone marrow, and a low risk of transformation to acute myeloid leukaemia.[4] This group of patients has a favourable prognosis compared with groups who have other chromosomal aberrations.[5]

Herein, we present for educational purposes, the image obtained from a bone marrow sample and the karyotype of a MDS case with 5q- syndrome seen at the Haematology and Haemotherapy Centre of the State University of Campinas. The patient, an 82-year-old woman, presented mild macrocytic anaemia (Hb: 9.1g/dL, Ht: 25.8%, MCV: 94.0fL, MCH: 29.9pg, reticulocyte: $3.0x10^6$/L) with normal leukocyte ($4.6x10^9$/L) and platelet ($316.0x10^9$/L) counts.

The bone marrow cytologic and histologic evaluations showed hypercellularity of the erythroid, granulocytic and megakaryoblastic lineages, with atypical erythroblasts and a great number of small and hypolobulated megakaryocytes (Figure 1). The karyotype was 46,XX,del(5)(q13q33) (Figure 2).

The patient has been seen in our service for two years, without red blood cell transfusions or transformation to acute leukaemia.

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