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Cytogenetic abnormalities occurs in almost half of myelodysplastic syndrome (MDS) patients and are considered to be the most important prognostic factor for survival, so they have been included in the International Prognostic Scoring System (IPSS) since 2007. Complex karyotype (CK) is defined as more than or equal to 3 independent chromosomal abnormalities at the same metaphase and is an independent poor prognostic factor, usually associated with a shorter median overall survival and propensity toward malignant transformation. From January 2008 through June 2022, 1744 patients with suspected or diagnosed hematological neoplasms (including MDS/AML) who had a bone marrow evaluation and concurrent conventional karyotype were identified. Karyotyping was successfully performed on 1167 (67%) of 1744 patients. In total, 914 (54%) with an adequate karyotypic analysis carried out a diagnosis of myelodysplastic syndrome. Among the cases, 9,7% were classified as complex karyotype. Del(5q)/-5 was identified in 52% of patients with complex karyotype, followed by chromosome 11 and chromosome 7 as well as chromosome 17 abnormalities, accounting for 38%, 23% and 23%, respectively. Besides, complex karyotype patients with translocations and derivation involvements, accounted for 28% and 14%, respectively. It is likely that pathogenetic mechanisms in del(5q) may involve hemizygous mutations or haploinsufficiency and be modified by additional somatic lesions affecting genes on other chromosomes. This report shows that the presence of deletion 5q may be the first step to the development of complex karyotype and is a determinant key of the genetic and genomic complexity and clonal hierarchy in MDS with 5q abnormalities.

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HOW IMPORTANT IS THROMBOCYTOSIS FOR PROGNOSIS OF MDS WITH 3Q21Q26 SYNDROME?

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The structural changes involving the long arm of chromosome 3 at bands 3q21 and 3q26.2 in the form of inversion are named paracentric inversion inv(3)(q21q26.2) and in myeloid neoplasms have long been recognized, but are rare. The 3q21q26 syndrome usually occurs in a high-risk myelodysplastic syndrome (MDS) or the setting of acute myeloid leukemia (AML) and is most commonly reported as inv(3)(q21q26.2). Myeloid neoplasms with inv(3) are rare disorders with an incidence of 1% in MDS and AML. Thus, this report aims to show a patient with MDS and high platelets count who presented inv(3)(q21q26.2). A 72-year-old woman looked for medical attention due to fatigue and weakness. The patient reported a history of smoking for 41 years and denied any exposure to toxic agents. At physical examination, only pale was detected. A complete blood count revealed hemoglobin 7g/dL, MCV = 94 fL, leukocytes 5,600/mm3, neutrophils 2,242/mm3 and thrombocytosis with a platelet count of 514,000/mm3. Bone marrow aspirate showed dyserythropoiesis in 30% of cells and 6,5% blasts. The bone marrow cytogenetic analysis showed 46,XX,inv(3)(q21q26.2)[16]/46,XX[4]. The diagnosis of MDS with excess blasts - 1 was established according to the 2016 World Health Organization classification and International Prognostic Scoring System was very high. While waiting for beginning treatment, the patient died of respiratory failure due to COVID-19. Myelodysplastic syndrome with inv(3)(q21q26.2) is a rare aggressive disorder that occurs in less than 1% of all MDS cases and has been associated with a poor outcome: chemoresistance, high risk of leukemic transformation and short survival. Our case showed thrombocytosis with a platelet count of 514,000/mm3. The incidence of thrombocytosis in MDS has been reported in 8% of cases with platelets $> 400 \times 109$/L. The major report which evaluated thrombocytosis in MDS studied 2,042 cases, detecting high platelets count in 5% of cases (102/2,042). It appears that thrombocytosis does not adequately capture the aggressive nature of inv(3)(q21q26.2) in MDS but still plays an important role in the pathogenesis of this heterogeneous and dynamic disease. Our patient reported herein showed dyserythropoiesis in 30% of cells and 6,5% blasts, but nothing was detected regarding megakaryocytic lineage. Our patient died very soon after diagnosis due to viral infection. Thrombocytosis is an unusual clinical feature in MDS associated with inv(3)(q21q26.2) and the unfavorable prognosis of inv(3) is independent of thrombocytosis.

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CASO CLÍNICO DE SMD RELACIONADA A QUIMIOTERAPIA PRÉVIA COM INDEPENDÊNCIA TRANSFUSIONAL APÓS TERAPIA DE SUPORTE ASSOCIADA A ELTROMBOPAGUE OLAMINA

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