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Refractory anemia with ring sideroblasts, with JAK2 and SF3B1 mutations without thrombocytosis: A case report and review of literature

JAK2 mutations are rare in refractory anemia with ring sideroblasts (RARS). We present the case of an 83-year-old female who was presented with anemia and no evidence of cytosis. Blood count revealed the following: WBC 4.9 K/uL, Hgb 9.7 g/dL and platelets 174 K/uL. The bone marrow was variably cellular and showed trilineage hematopoiesis. Erythroid precursors appeared normal in number; however, numerous ring sideroblasts were identified. There were no karyotypic or FISH abnormalities. Molecular studies using next generation sequencing technology showed both JAK2 and SF3B1 mutations. These findings were most consistent with a Myelodysplastic syndrome, RARS. SF3B1 mutations are associated with a favorable prognosis in MDS, and are highly predictive for the presence of ring sideroblasts. The presence of the JAK2 mutation in this patient is unusual. JAK2 mutation usually leads to cytokine hypersensitivity and cytokine-independent growth of hematopoietic cells resulting in uncontrolled proliferation and a myeloproliferative phenotype. JAK2 mutation is rare in myelodysplastic syndromes such as RARS. However, the presence of both JAK2 and SF3B1 mutations has been seen in cases of myelodysplastic/myeloproliferative neoplasms (MDS/MPN), such as RARS associated with marked thrombocytosis (RARS-T). The current case had normal platelet count, and therefore did not satisfy the criteria for RARS-T. The possibility that this patient would later develop an MDS/MPN cannot be excluded. The reason that some MDS patients with JAK2 mutation do not develop a myeloproliferative component is unknown. Further studies are needed to determine the role of JAK2 mutation in these MDS patients.

Biography

Cynthia Lorenzo has completed her MD from Stanford University School of Medicine. She is a Senior Hematopathologist at Genoptix Medical Laboratory, USA. She has her publications in many reputed journals.

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