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Myelodysplastic Syndromes with Ring Sideroblasts: a Monocentric Study in the South of Tunisia

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BACKGROUND

- ✓ Myelodysplastic syndromes with ring sideroblasts (MDS-RS) are clonal disorders of pluripotent stem cells often associated with the presence of a mutation in the SF3B1 splicing factor.
- ✓ They are rare hematological disorders of the elderly, with a slight female predominance, and are most often idiopathic.
- ✓ The aim of our work is to study a patient cohort in southern Tunisia.

MATERIALS & METHODS

Hematology laboratory of the Habib Bourguiba university hospital in Sfax.

12 years :
January 2006- December 2017

All cases diagnosed with MDS-RS based on the 2016 WHO classification of MDS

Results

12 patients

AGE Median age 64 year

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- The anemic syndrome was ubiquitous at diagnosis, with constant bone marrow dyserythropoiesis.
- Bone marrow dysplasia was monolineage in 8 patients, and the median percentage of ring sideroblasts was 27%.
- Ferritinemia and folate/vitamin B12 levels, measured in 7 and 9 patients respectively, were normal or increased, thus ruling out deficiency anemia.
- The bone marrow karyotype analysis performed on 10 patients came back without abnormalities.
- In our cohort, the patients were at low risk of progression according to the International Prognostic Scoring System, and the therapeutic approach was therefore to correct the cytopenias.

DISCUSSION

- Ring sideroblasts (RS) are erythroid precursors with abnormal perinuclear mitochondrial iron accumulation.
- In 2011, with the advent of next generation sequencing (NGS) technology, somatic spliceosome component mutations (SF3B1-2q33.1, SRSF2-17q25.1, U2AF1-21q22.3, ZRSR2-Xp22.1, SF3A1-22q12.2 and U2AF2-19q13.42) were first described in patients with MDS. Amongst these, SF3B1 mutations were most common in patients with MDS-RS.
- SF3B1 mutations can be seen in ~80% of patients with MDS-RS-SLD, ~40% of patients with MDS-RS-MLD, with the percentage of BMRS often correlating directly with the SF3B1 mutant variant allele frequency burden.
- SF3B1-mutated MDS are a unique MDS subtype with favorable outcomes.

CONCLUSION

Myelodysplastic syndrome with ring sideroblasts remains a sparsely-studied disease, both in our country and worldwide. A national multicentric study will provide further support for our findings.