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CHARACTERIZATION OF CHROMOSOMAL ABNORMALITIES ASSOCIATED WITH POOR PROGNOSIS IN A GROUP OF MEXICAN MESTIZO PATIENTS AFFECTED WITH MYELODYSPLASTIC SYNDROME

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INTRODUCTION

Myelodysplastic syndrome (MDS) is an heterogeneous group of clonal disorders characterized by abnormal cellular maturation, cytopenia and elevated risk of progression to acute myeloid leukemia (AML). Chromosome analysis has a crucial role in evaluate the prognosis of MDS patients and has been a key item of the International Prognostic Scoring System Revised (IPSS-R). Half of MDS patients have at least one chromosome abnormality identified by karyotype. The poor prognosis category, based on the IPSS-R model, includes patients with complex karyotypes (with more than 3 chromosomal abnormalities) and it is normally associated with resistance to chemotherapy. The meaning of the abnormalities identified in this category are still debatable.

AIM

Characterize the cytogenetic abnormalities in complex karyotypes present in a Mexican MDS population using standard G-band chromosome analysis.

METHOD

Design: The study was performed in a Mestizo Mexican Population diagnosticated with MDS in a fourteen year period (2010-2024).

Setting: All cases were referred for cytogenetic studies to Mendel Laboratory, a private institution, the cases were referred from institutions all over Mexico and selected based on the diagnosis referred from the physician.

Participants: A total of 1223 cases were selected based on the diagnosis and samples from each case were evaluated and processed according to standard protocols.

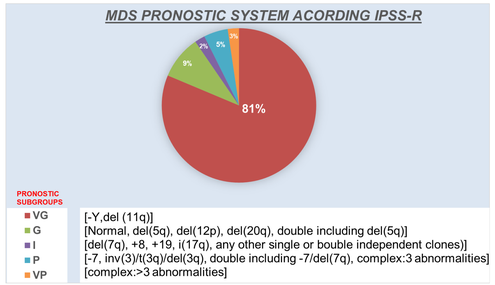
Interventions: Samples of each case (bone marrow or peripheral blood) were processed for chromosome analysis and classified according to IPSS-R.

RESULTS

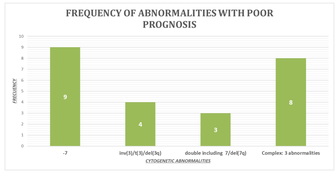
A total of 1223 cases with MDS diagnosis were included in this study.

Out of them 901 (81%) patients were classified as good prognosis according to IPSS-R. 57 (5%) showed a complex karyotype consistent with poor prognosis.

Besides chromosome 7 and 5 abnormalities, aneuploidy of chromosome 17 and 21 were more prevalent. Structural abnormalities in chromosomes 1, 3, 11 and 12 were frequently identified as part of the cytogenetic analysis.

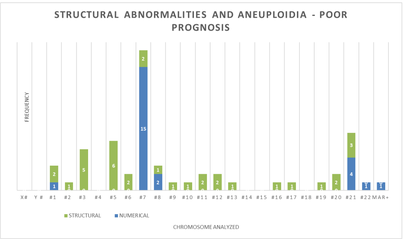


Distribution of the risk group according to the IPSS-R classification in patients with myelodysplastic syndrome

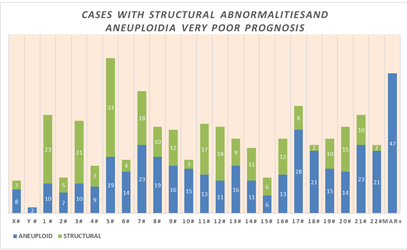


The frequency is shown according to the subcategories of myelodysplastic syndrome with poor prognosis. Which the main abnormality is the aneuploidy of chromosome 7

The distribution of abnormalities associated with the poor prognosis category is shown. These are shown by chromosome, separating aneuploidies and structural abnormalities, which were counted for each observation presented in the analysis. The chromosomes with the most alterations were 7, 21, 5 and 3.



The distribution of abnormalities associated with the very poor prognosis category is shown. These are shown by chromosome, separating aneuploidies and structural abnormalities, which were counted for each observation presented in the analysis. The distribution of abnormalities is more heterogeneous, with the main chromosomes affected being 5, 7, 17, 1, 21 and 3. It is important to highlight the presence of marker chromosomes in this risk classification.



CONCLUSIONS

Cytogenetics abnormalities have implications in the prognosis in MDS. Complex karyotypes are associated with poor prognosis and they are usually heterogeneous. Here we describe a panoramic view of the diversity of cytogenetic leukemic abnormalities associated with MDS. The expected results for this study was the prevalence of chromosome 5 and 7 abnormalities in complex karyotypes. However , the distribution of abnormalities is more heterogeneous, with the main chromosomes affected being 5, 7, 17, 1, 21 and 3. It is important to highlight the presence of marker chromosomes in this risk classification.

REFERENCES

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