Clinical Characteristics and Mutational Profile of Patients with Myelodysplastic Syndromes and Chronic Myelomonocytic Leukemia.

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Background

Myelodysplastic syndromes (MDS) and chronic myelomonocytic leukemia (CMML) are heterogeneous myeloid malignant clonal disorders. Next-generation sequencing (NGS) is a tool that allows the detection of useful characteristics for the diagnosis and prognosis of myeloid pathologies. An association between certain mutations and clinical characteristics has recently been proposed and predictive models of survival.

Objective

Describe clinical characteristics and their association with mutational profiles in adult patients with MDS and CMML diagnosis. Assess molecular risk models to predict outcomes.

Materials & Methods

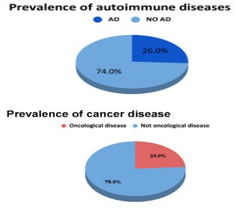
Retrospective cohort study, including adult patients diagnosed with MDS and CMML from two highly complex private centers in Latin America.

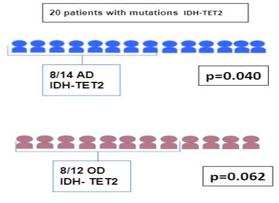
Results

55 patients were included. In table 1 patient characteristics. Karyotype abnormalities were found in 16 patients (29%), being the alterations in chromosome 8 (8/55), 7 (7/55) and 5 (6/55) the most frequent. Adverse Risk mutations were found in more than 50% (21/39) of patients with good cytogenetic risk. Of the 36 low/very low risk patients according to IPSS-R 4/36 (11%) and 6/36 (17%) were categorized at high and intermediate risk respectively with the MIPSS-Scale.

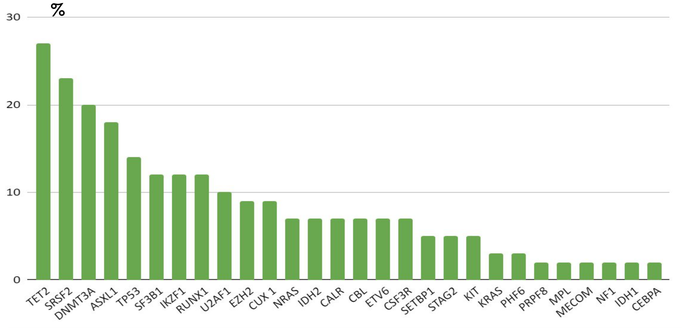
Table 1. Patients characteristics. N: 55

Female N (%)	23 (41)	**Mutations NGS N**	52 (91)
Age median (IQR)	55 (31-87)	**Number of mutations**	3 (0-6 IQR)
IPSS-R N (%)		**Karyotype**	**Abnormalities**
- Very Low	11 (20)	chromosome 8	8/55 (15)
- Low	25 (46)	chromosome 7	7/55 (13)
- Intermediate	7 (13)	chromosome 5	6/55 (11)
- High	5 (7)		
- Very High	7 (13)		



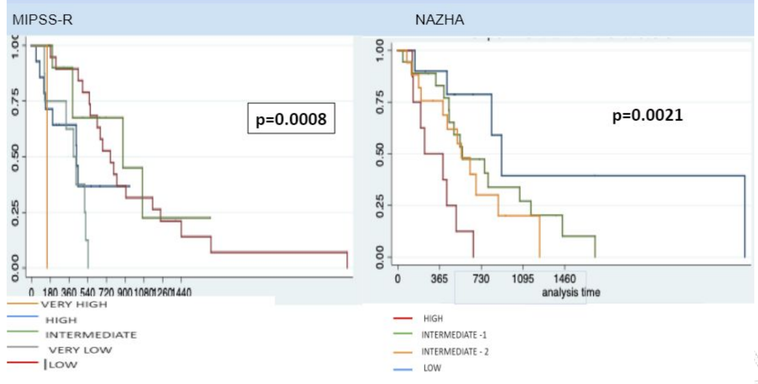


Patients with *TET/IDH* mutations had significantly higher cancer rates (p=0.040) and a higher trend of IE (p=0.062).



91% presented at least one somatic mutation by NGS. *TET2, SRSF2, DNMT3A, ASXL1* and *TP53* were the most frequently reported mutations, in 15, 13, 11, 10, and 8 patients respectively.

Overall survival according to molecular scales



The median OS for the entire group was 1.5 years, and at 2 years it was 27%.
The Nazha stratification scale and the MIPSS-R were good predictors of OS.

Discussion

Mutations in TET/IDH are a special group of interest with potential clinical implications. The molecular risk scales of Nazha and MIPSS-R were useful tools to predict OS.

References

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