Mutation Profiling and Characteristics of Hypoplastic MDS

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Background

Myelodysplastic syndromes (MDS) are heterogenous spectrum of myeloid neoplasms. A subset of patients with MDS have hypoplastic marrow (h-MDS) and is being recognized more as a separate entity in the upcoming 2022 WHO classification.

Objective

We aim to study characteristics and the molecular profiling of hypoplastic (h-MDS) in comparison with normo-/hypercellular MDS (n-MDS).

Methods

This was a retrospective analysis of all patients diagnosed with MDS from Moffitt Cancer Center MDS database. Patients were divided into two groups: those with h-MDS, defined as cellularity less than 30%, and those with n-MDS, cellularity greater than 30%. Individuals with MDS/MPN were excluded.

Conclusion

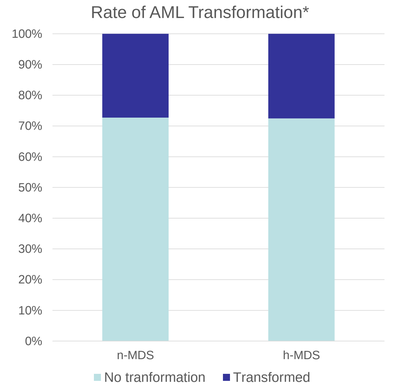
h-MDS accounted for 10% of MDS cases. The OS for h-MDS is better than n-MDS, which tended to have more frequency of splicing and epigenetic somatic mutations.

Results

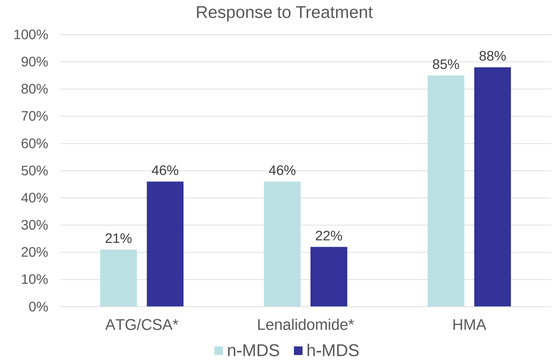
	Normocellular MDS (n-MDS)	Hypoplastic MDS (h-MDS)
Total number of patients 4,037*	3,634 (90%)	403 (10%)
Median age at diagnosis*	68	65
Female gender*	35%	41%
Mean blast percentage	5.27	5.3
Mean ANC (10*3/µL)*	2,220	1,4270
Mean platelet count (10*3/µL)*	138,000	116,000
Platelet transfusion*	23%	30%
t-MDS*	20%	29%
MDS-RS*	16%	4%
R-IPSS > 3*	36%	36%

t-MDS: Therapy related MDS. MDS-RS: MDS with ringed sidroblasts. ANC: Absolute neutrophil count.
*Statistically significant P value <0.05
Note: There was no difference between both groups in terms of IPSS-R, LGL PNH clones, trisomy 8 and history of autoimmune diseases.

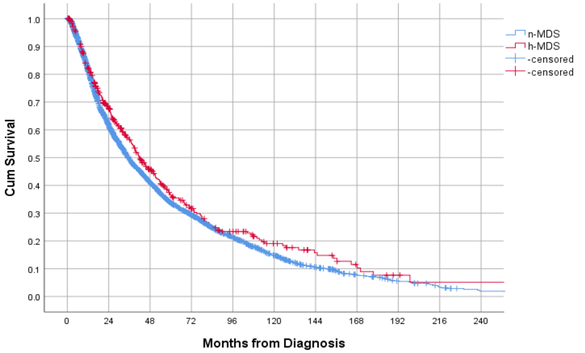
Mutations	Normocellular MDS (n-MDS)	Hypoplastic MDS (h-MDS)
SF3B1 (*P*<0.005)	19%	7%
SRSF2 (*P*<0.005)	13%	5%
TET2 (*P*=0.004)	23%	14%
ASXL1 (*P*=0.02)	21%	14%



*P value=0.9



*Statistically significant P value <0.05



Median overall survival (OS) for h-MDS was 41.6 months compared to 35 months in patients with n-MDS (*P*=0.05). The HR for OS was 0.88 (*P* =0.057) adjusted for IPSS-R.

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