SOHO ANNUAL MEETING 2024

UNVEILING THE SPECTRUM OF FISH ABNORMALITIES IN A DECADE LONG RETROSPECTIVE STUDY OF MULTIPLE MYELOMA : INSIGHTS FROM AN UNDERREPRESENTED POPULATION

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INTRODUCTION

Fluorescent in situ hybridization (FISH) is a powerful technique for detecting cytogenetic (CG) abnormalities in Multiple Myeloma (MM). Despite their diagnostic and prognostic significance, data on their frequency are scarce. Our study aims to enrich the data on their frequency in an underrepresented population.

AIM

- **Characterize the Frequency and Types of CG Abnormalities in MM patients.**
- **Correlate CG Findings with Demographic Data and enhance understanding of MM Heterogeneity.**

METHOD

The study conducted a retrospective analysis of MM patients confirmed by biopsy at a cancer center in Brooklyn from 2013 to 2024. Inclusion criteria included bone marrow biopsy reports with FISH analysis of patients with MM. Comprehensive demographic information, FISH analysis outcomes, and specific abnormalities were diligently collected and analyzed

RESULTS

A total of 135 patient records were reviewed, of which 97 (71.8%) were confirmed as MM cases, with a median age of 69 years (range 36–90). Among these patients, 71 (52.5%) were male, and 64 (47.4%) were female. However, only 49 patients met the study's inclusion criteria and had FISH analysis data. Of these, 79.5% were African American, with 25 (51%) females and 24 (49%) males. Abnormal FISH findings were observed in 36 patients, with multiple abnormalities detected in 25 patients.

Table 1: Risk Stratification of the Multiple Myeloma patients based on FISH analysis.

Sr No	Mutations	Details
1	**High-Risk**	Mutations: P53 mutation, 17p deletion, gain of 1q, del(1p), t(14;16), t(14;20)
2	**Intermediate-Risk**	Mutations: t(4;14), gain 1q25
3	**Standard-Risk**	Mutations: isolated monosomies, isolated hyperploidy, t(11;14), t(6;14)
4	**Low-Risk**	Mutations: Normal FISH

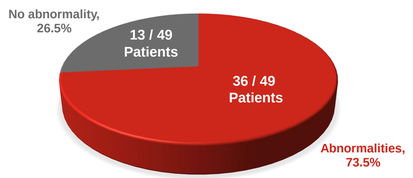


Figure 1: Proportion of Cases with Cytogenetic Abnormalities vs No Abnormalities as found in the FISH analysis of Multiple Myeloma Patients.

Table 2: Distribution of Multiple Cytogenetic Abnormalities found in the Multiple Myeloma patients based on the FISH analysis.

Sr No	Category	No. of Patients	%	Most Common
1	**Patients with abnormalities**	**36**	**73.5%**	
1.1	Hyperploidy	18	36.7%	Chromosome 9,15 and 11
1.2	Translocation	16	32.6%	CCND1 and 1q21
1.3	Gene gain/amplification	13	26.5%	IGH/CCND1 (t11:14)
1.4	Hypoploidy	10	20.4%	Monosomy 13
1.5	Deletions	8	16.3%	Tp53/17p
2	**Patients with no abnormalities**	**13**	**26.5%**	-

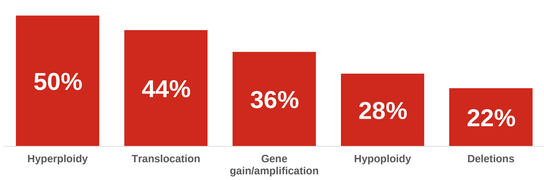


Figure 2: Distribution of Cytogenetic Abnormalities in Multiple Myeloma Cases in the study based on FISH reports.

CONCLUSIONS

In the study cohort, 44.44% of patients had high-risk cytogenetics, 13.88% were intermediate-risk, and 41.66% were standard-risk. Hyperploidy was the most frequent abnormality, while translocations t(14;16) and t(4;14) were rare. The prevalence of multiple cytogenetic abnormalities underscores the heterogeneity within MM, offering valuable insights for future research

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ACKNOWLEDGEMENTS

Our gratitude to Brooklyn Cancer Care for providing the necessary facilities and resources. Special thanks to Dr. Khillan for his mentorship and Bryana Mojica for her assistance.

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