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Conference Abstract A1

Opportunities for Genetic Stratification in Personalized Treatment: A Case Study of Multiple Myeloma

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Introduction

Multiple myeloma (MM) is a heterogeneous disease in which patient survival and treatment response are closely linked to genetic mutations. Recent studies show that identifying genetic biomarkers enables personalized treatment for patients [1]. In particular, mutations such as translocation t(11;14)(q13;q32), deletion 17p (P53), and duplication 1q (CKS1B) are considered key genetic markers for therapy selection [2]. This article analyzes the frequency and clinical significance of several genetic abnormalities.

This is the first study in Azerbaijan devoted to genetic stratification in MM patients, analyzing the frequency and clinical relevance of specific genetic abnormalities. Some rare alterations (hypodiploidy 13 and 14) were also recorded, highlighting regional features of the disease.

Methodology

The study was conducted at the National Center of Hematology and Transfusion of the Ministry of Health of the Republic of Azerbaijan between 2020 and 2023 and included 28 patients diagnosed with MM. Diagnosis was based on the International Myeloma Working Group (IMWG, 2014) criteria [1]. For genetic analyses, fluorescence in situ hybridization (FISH) and next-generation sequencing (NGS) methods were used. The obtained data were analyzed retrospectively, and mutation frequencies were determined.

Results

The study revealed that translocation t(11;14)(q13;q32) was found in 21.4% of patients (6 individuals) and was the most frequent genetic abnormality. This translocation is associated with sensitivity to BCL-2 inhibitors [3]. Deletion 17p (P53) and duplication 1q (CKS1B) were detected in 10.7% of patients, indicating a more aggressive course of the disease [4]. These results confirm the impact of genetic mutations on prognosis and treatment selection.

The following table summarizes the main mutations and their frequency (see Figure 1). Results demonstrate that genetic mutations are heterogeneously distributed among patients, and different mutations have varying prognostic significance.

Mutation	Number of patients (n)	Percentage (%)
t(11;14)(q13;q32)	6	21.4
Trisomy 11	3	10.7
17p (P53) deletion	3	10.7
1q CKS1B duplication	3	10.7
1p CDKN2C deletion	3	10.7
13q deletion	2	7.1
13 and 17 hypodiploidy	1	3.5
Trisomy 5	1	3.5
Monosomy 13	1	3.5
Tetrasomy 14	1	3.5
Trisomy 17	1	3.5

Conclusion

This study confirms the important role of genetic mutations in personalized treatment and prognostic assessment of MM patients. Risk group determination based on identified biomarkers and development of individualized therapy strategies may contribute to improved patient survival.

Future research should explore in greater depth the relationship between genetic mutations and treatment response, as well as the efficacy of novel targeted therapies.