Role of Immunohistochemistry in Nucleophosmin 1 (NPM1) Mutated Acute Myeloid Leukemia (AML)

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Abstract

Background: Acute myeloid leukemia (AML) is characterized by the abnormal proliferation of myeloid blasts in the blood, bone marrow, and other tissues. Nucleophosmin 1 (NPM1) mutations are the most frequent genetic alteration in AML that are now considered diagnostic for AML even at lower blast count thresholds than traditionally required.

Objective: Present study aims to analyze the role of immunohistochemistry in detecting NPM1 mutated cases of AML and to compare it with molecular studies which is a gold standard for the detection of NPM1 gene mutation.

Methodology: 50 patients with the diagnosis of AML were enrolled in the study. Bone marrow biopsy samples are subjected to IHC using anti-NPM1 antibody. Cases showing both nuclear and cytoplasmic staining (predominantly cytoplasmic) are considered positive for NPM1 mutation. The IHC results were correlated with molecular findings obtained through RT-PCR or NGS.

Results: Preliminary analysis of 20 cases showed that all NPM1 mutation–positive cases demonstrated both nuclear and cytoplasmic staining on IHC, with predominant cytoplasmic localization. The IHC results showed complete concordance with molecular findings, indicating high sensitivity and specificity.

Conclusion: Immunohistochemistry for NPM1 showing combined nuclear and cytoplasmic (predominantly cytoplasmic) staining serves as a reliable, rapid, and cost-effective diagnostic tool for identifying NPM1-mutated AML. The strong concordance with molecular testing supports its use as a practical screening and adjunct diagnostic method, especially in routine hematopathology and resource-limited laboratories.

Keywords

Acute myeloid leukemia, Nucleophosmin 1, Immunohistochemistry, Molecular diagnosis, NPM1 mutation.