A RARE CASE OF ACUTE PROMYELOCYTIC LEUKEMIA WITH PML::RARA FUSION PRESETING WITH INITIAL ATYPICAL IMMUNOPHENOTYPIC FEATURES LEADING TO DIAGNOSTIC COMPLEXITY

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Background: Acute promyelocytic leukemia (APL) is a distinct subtype of acute myeloid leukemia characterized by the PML::RARA fusion resulting from t(15;17)(q22;q21). Although molecular confirmation is considered diagnostic, atypical immunophenotypic and morphologic findings may complicate the initial diagnosis and delay appropriate treatment.

Purpose: To present a rare case of APL with confirmed PML::RARA fusion and initial atypical immunophenotypic features, highlighting the importance of an integrated diagnostic approach.

Methods: Conventional cytogenetic analysis, fluorescence in situ hybridization (FISH), reverse transcription polymerase chain reaction (RT-PCR), and next-generation sequencing (NGS) were performed. Molecular analyses included PML::RARA, BCR::ABL1, RUNX1::RUNX1T1, NPM1, FLT3, and CEBPA. Bone marrow morphology and multiparametric flow cytometric immunophenotyping were also evaluated. Results A female patient presented with suspected acute leukemia. RT-PCR confirmed the PML::RARA fusion, while FISH and conventional karyotyping demonstrated 46,XX,t(15;17)(q22;q21). No additional recurrent AML-associated genetic abnormalities were detected, whereas a CEBPA promoter variant (c.-36C>T) was identified. Flow cytometry demonstrated a myeloid blast population expressing CD13, CD15, CD16, CD24, CD33, CD36, CD56, and myeloperoxidase (MPO), with absent CD34 and CD117 expression. Bone marrow biopsy revealed hypercellular marrow with diffuse infiltration by abnormal promyelocytes and strong MPO positivity. Despite the initial atypical immunophenotypic findings, integration of molecular, cytogenetic, immunophenotypic, and morphologic results established the diagnosis of APL.

Conclusions: This case demonstrates that APL may initially present with atypical immunophenotypic features despite definitive PML::RARA confirmation. Comprehensive integration of molecular, cytogenetic, flow cytometric, and morphologic findings is essential for timely diagnosis and appropriate clinical management.