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617.ACUTE MYELOID LEUKEMIAS: BIOMARKERS, MOLECULAR MARKERS AND MINIMAL RESIDUAL DISEASE IN DIAGNOSIS AND PROGNOSIS

Diagnostic Challenges in the Leukemia Phase of Blastic Plasmacytoid Dendritic Cell Neoplasm without Skin Involvement: A Clinical and Pathological Study

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Objective: To improve the accurate diagnosis and treatment of blastic plasmacytoid dendritic cell neoplasm (BPDCN) without skin involvement. **Methods**: One case of BPDCN with skin involvement progressing to the leukemia phase was used as a positive reference for bone marrow specimens, and a comparative study was conducted on one case of leukemia phase of BPDCN without skin involvement. **Results**: The male control was 70 years old and presented with a mass on the right forearm skin. Pathological examination indicated BPDCN. Only one cycle of decitabine + CAG was given and 5 months later, the disease progressed to the leukemia phase. Tumor cells expressed HLA-DR, CD123, CD4, CD56, CD7, CD36, TCL1 and CD304. NGS analysis of bone marrow tumor indicated ASXL1, IDH2, and TP53 mutations. The patient survived only 6 months during follow-up. Another female patient was 73 years old and was initially diagnosed with "T lymphoblastic leukemia" based on partial immunophenotyping results for 81.5% blast cells and the morphology of blast cells showing tailing phenomena. Further immunophenotyping showed that blast cells expressed CD56, HLA-DR, and CD304, partially expressed CD4, CD33, and CD117, and did not express CD34, CD13, CD3, CD5, CD7, CD8, CD10, CD11b, CD14, CD15, CD16, CD19, CD20, CD64, CD303 or MPO. Additional immunohistochemistry of bone marrow biopsy showed that CD43 was negative, TdT was mostly positive, CD4 was slightly positive, CD56 was mostly positive, CD123 was positive, CD117 was slightly positive, and lysozyme was partially weakly positive. The above results implied that after excluding BPDCN, acute myeloid leukemia subtype M5 should be considered. According to the diagnostic criteria of BPDCN in WHO 5th edition hematologic lymphoma, two scenarios can be established for diagnosing BPDCN: (1) CD4 and/or CD56 are positive and CD123 and one of pDC markers (TCL1, CD303, CD304) are expressed. (2) If any three pDC markers are expressed (CD123, TCL1, CD303, CD304), and all expected negative markers (CD3, CD14, CD19, CD34, Lysozyme, Myeloperoxidase) are negative, So the final diagnosis of BPDCN can be established according the former creteria. The latter case had ASXL1 and TET2 mutations detected by NGS analysis, achieved complete remission after receiving venetoclax + azacitidine for one cycle, followed by intermittent venoclax +demethylation agent or low-dose chemotherapy maintenance treatment and live for more than two years now. **Conclusion**:BPDCN without skin lesions is clinically rare, and its diagnosis is challenging. Comprehensive immunophenotyping and cautious interpretation of immunohistochemistry results such as dim lysozyme expression are crucial. Venetoclax-containing regimens have shown promising therapeutic effects.

Disclosures No relevant conflicts of interest to declare.

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