Original Research

HEPATOSPLENIC AB T-CELL LYMPHOMA WITH ISOCHROMOSOME 7Q AND TRISOMY 8: A RARE DIAGNOSTIC CHALLENGE WITH CYTOGENETIC CONFIRMATION ASSOCIATED WITH RAPID DISEASE PROGRESSION AND HIGH MORTALITY

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ABSTRACT

Background: An uncommon and extremely aggressive form of peripheral T-cell lymphoma, HSTCL (Hepatosplenic T-Cell Lymphoma) is distinguished by extranodal involvement of the liver, spleen, and bone marrow. It is linked to a poor prognosis and makes up less than 5% of peripheral T-cell lymphomas.

Case Presentation: A 33-year-old male presented with a three-month history of fever, recurrent epistaxis, pancytopenia, and massive splenomegaly. Initial bone marrow examination demonstrated dysplastic changes suggestive of myelodysplastic syndrome. Flow cytometric immunophenotyping subsequently identified a clonal CD8-positive αβ T-cell population expressing CD2, CD3, CD7, CD16, CD38, and CD94 with TRBC1 restriction. Cytogenetic analysis revealed isochromosome 7q and trisomy 8, characteristic abnormalities associated with hepatosplenic T-cell lymphoma. The patient received CHOP-E-based chemotherapy and multiple transfusions for severe cytopenias. Despite prompt initiation of timely treatment, he succumbed to progressive disease, again highlighting the aggressive biology and poor prognosis of the disease .

Conclusion: HSTCL should be considered in young patients presenting with unexplained pancytopenia, hepatosplenomegaly, and absence of significant lymphadenopathy. Early utilization of flow cytometry and cytogenetic studies may facilitate timely diagnosis of this aggressive malignancy.

Keywords: Hepatosplenic T-Cell Lymphoma, Pancytopenia, Splenomegaly, Isochromosome 7q, Trisomy 8, Peripheral T-Cell Lymphoma, Cytogenetics.

INTRODUCTION

A rare subtype of PTCL (Peripheral T-cell Lymphoma), HSTCL (Hepatosplenic T-Cell Lymphoma) was originally characterized in 1981 and was added to the new European American Lymphoma Classification in 1994 as a provisional entity γδ hepatosplenic T-cell lymphoma.Large sinusoidal infiltration of medium-sized cytotoxic T-cells in the liver, spleen, and bone marrow is a characteristic of HSTCL, [1] HSTCL accounts for less than 1% of all non-Hodgkin lymphomas and less than 5% of peripheral T-cell lymphomas, making it one of the rarest mature T-cell neoplasms. Median overall survival reported in most series ranges from 8–16 months despite intensive chemotherapy.[2]

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Case Report

A 33-year-old male with no comorbidities, presented with fever since 3 months, and history of epistaxis, which was severe enough to warrant blood transfusion. There was modest hepatomegaly and significant splenomegaly that reached the right iliac fossa. Laboratory investigations revealed pancytopenia. Ultrasound of the abdomen revealed hepatosplenomegaly and no lymphadenopathy. Dimorphic anaemia of severe degree with

leucopenia and thrombocytopenia. The patient was evaluated for fever with malaria, dengue, and blood culture, which were within normal limits. HIV/HCV/HBSAG was negative. HRCT thorax did not show significant abnormalities. Echo showed mild pericardial effusion and was otherwise unremarkable.

Bone marrow biopsy showed erythroid hyperplasia with an increase in the immature precursor forms. ALIP (Abnormal Localization of Immature Precursors) noted with the presence of immature forms in the central parts of marrow megakaryocytes is increased in number with multi nucleation and nuclear hyplobation depicting features of dysmegakaryopoiesis. The patient's young age, massive splenomegaly, and severe constitutional symptoms were atypical for primary MDS and prompted further immunophenotypic evaluation. Serum iron studies were within normal limits. The MPN panel showed pathogenic mutation in the PHF 6 gene.

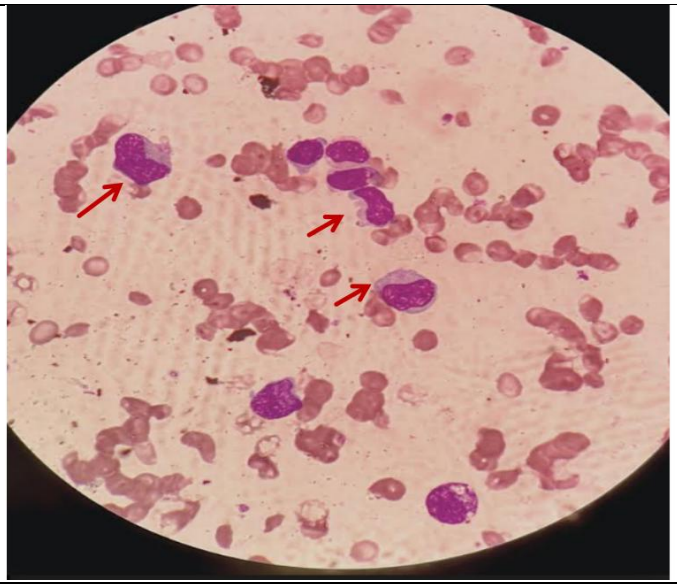

Figure 1: Abnormal Lymphoid Cells in the Bone Marrow Aspirate

Flow cytometric immunophenotyping showed a population of CD8 (dim to moderate) positive T cells that expressed CD3, CD7 (moderate), CD2 (bright), CD16 (subset), CD38, CD94 and TCR-Alpha/Beta and showed clonality on TRBC1 assessment. These cells were negative for CD56, CD57, CD5, TCR Gamma delta, CD4 and other markers ofcell and myeloid differentiation.

In view of the lack of Large granular lymphocytes or prolymphocytes on the smear and the lack of significant evidence of liver involvement clinically, a peripheral T cell lymphoma was considered likely. CHOP-E (Cyclophosphamide, doxorubicin hydrochloride (hydroxydaunorubicin), vincristine sulphate (Oncovin), prednisone, and etoposide) was the chemotherapy protocol used to treat him. A few days later, the karyotyping report revealed that most metaphases had isochromosome 7q and trisomy 8, which are consistent primary, non-random. Hepatosplenic T alpha/beta lymphoma is linked to such chromosomal abnormalities. Patient had repeated transfusions in view of epistaxis and severe anemia and thrombocytopenia. Patient had received a total of 3 cycles of chemotherapy. He also continued to have repeated infections and shortly succumbed to the illness.

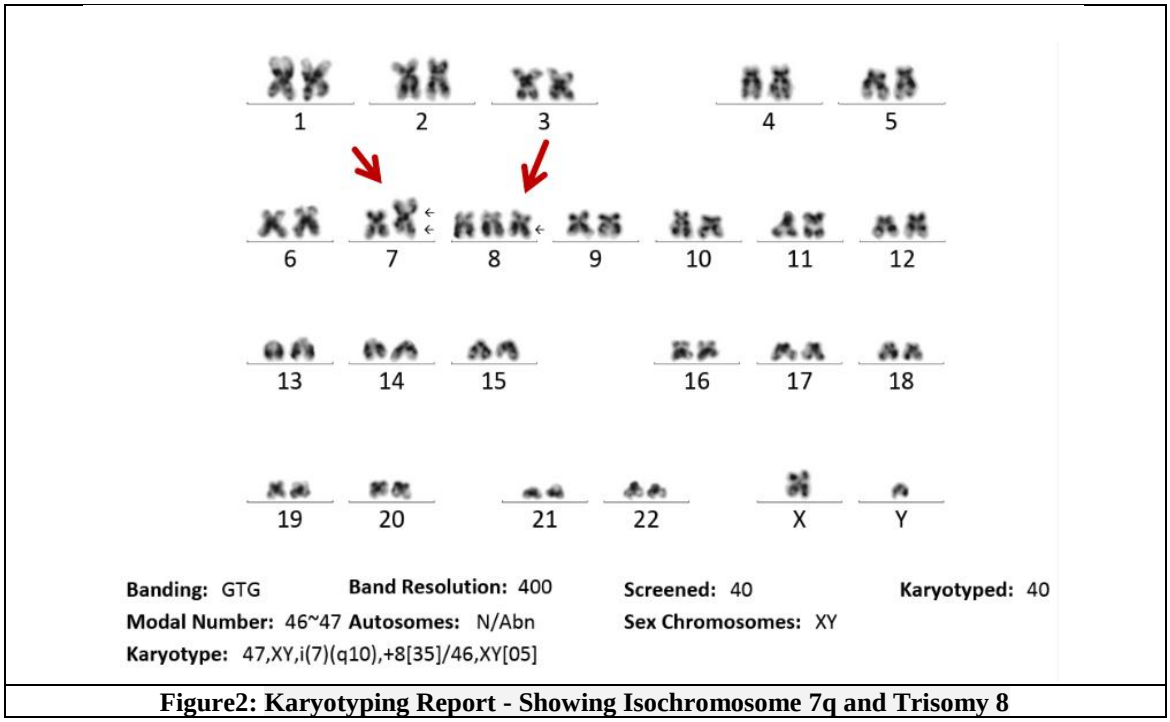

Figure2: Karyotyping Report - Showing Isochromosome 7q and Trisomy 8

DISCUSSION

The prevalence of this lymphoma is higher in young adults and teenagers. It is resistant to traditional chemotherapy regimens, leading to a rapidly progressive clinical course and a dismal prognosis.[2] About 20% of cases have an underlying history of immunological dysregulation or chronic immunosuppression.[2] HSTCL is classified based on the T-cell receptor (TCR) expressed by the malignant cells:. γδ (Gamma-Delta) HSTCL accounts for 80% of the cases . αβ (Alpha-Beta) HSTCL accounts for 20% of the cases. [2]

Constitutional symptoms, abdominal pain from splenomegaly or hepatomegaly, and, in more severe situations, jaundice are the usual presentation symptoms for patients. While lymphadenopathy is rarely present, cytopenias are prevalent. Almost invariably present, thrombocytopenia has been linked to the advancement of the disease. Flow cytometry can identify a small population of aberrant cells in approximately 50% of patients, despite lymphocytosis being an uncommon condition. Leukemic and blastic evolution have been described in advanced cases.[3]

The liver, spleen, and bone marrow are usually affected in a sinusoidal pattern by HSTCL. Approximately two thirds of individuals have bone marrow involvement upon diagnosis, and this number rises as the disease progresses. Neoplastic T cells can sometimes mimic blasts, and the marrow infiltrate is either sinusoidal or interstitial. As additional blastic cells are included, bone marrow infiltration may become more interstitial. CD3 immunohistochemical stain can be used to reveal the sinusoidal pattern of invasion.[4,5]

The most prevalent chromosomal abnormalities in HSTCL are trisomy 8 and chromosome 7q [i(7q)], which are found in up to 50% and 63% of cases, respectively, and commonly co-occur. It has been suggested that i(7q) is the primary genetic event and that trisomy 8 and 7q amplification are secondary genetic events acquired throughout development. Isochromosome 7q is regarded as the cytogenetic hallmark of HSTCL and is thought to represent an early pathogenic event. The coexistence of trisomy 8 further strengthens the diagnosis and has been reported in a substantial proportion of cases.[6,7]

This case is noteworthy for a number of reasons. Firstly, the lymphoma was of the less common αβ T-cell phenotype. Secondly, the first marrow morphology was very similar to myelodysplastic syndrome, which was a diagnostic pitfall. Thirdly, the cytogenetic analysis confirmed the presence of the characteristic combination of the isochromosome 7q and trisomy 8 as strong diagnostic confirmation. Finally, despite CHOP-E chemotherapy, rapid progression and death in this patient demonstrated an aggressive natural history of this disease.

Two single-institution studies reported results with cyclophosphamide, doxorubicin, vincristine, and prednisone (CHOP) or CHOP-like regimens. 21 cases of γδ HSTCL were described by Belhadj et al. 9 patients had a complete response (CR), and the overall response rate was 73%. Even then, only two patients remained alive at the time of the research, and the median survival duration was just 16

months, despite the early response. It's interesting to note that all patients' advancement involved the original areas of their disease, and recurrent thrombocytopenia was seen at that point.[8,9]

A history of immunocompromised, male sex, and failure to attain a CR were all considered poor prognostic factors. Many patients have transient responses to standard CHOP-based chemotherapy but rarely achieve durable remission. Current evidence indicates that intensive induction chemotherapy followed by allogeneic stem cell transplantation in first remission offers the best chance of long-term survival. For patients with post-transplant HSTCL, the prognosis is poorer.[2]

CONCLUSION

HSTCL is a unique and rare but very aggressive lymphoma where relapses occur frequently, the response to first treatments is subpar, and the median overall survival is short. It presents with hepatosplenomegaly, cytopenias, absence of significant lymphadenopathy and characteristic cytogenetic abnormalities including isochromosome 7q and trisomy 8. A high index of suspicion is essential as it may mimic bone marrow failure syndromes including myelodysplastic syndrome. Incorporation of flow cytometry and cytogenetic studies can aid early diagnosis. Outcomes remain poor despite treatment, highlighting the importance of early referral to specialized centers and stem cell transplantation whenever possible.[10]

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