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https://doi.org/10.1016/j.htct.2025.104003

13

UNEXPECTED FINDING IN THE PERIPHERAL BLOOD EVALUATION OF A HUMAN T-LYMPHOTROPIC VIRUS TYPE 1 (HTLV-1) SEROPOSITIVE PATIENT

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Objective: The aim of this study is to report the unexpected finding of a clonal T-cell population in a carrier of HTLV-1 virus and review the pertinent literature. **Materials and methods:** Case report and review of the literature. **Case report:** Male patient, 55 years old, asymptomatic carrier of the HTLV-1 virus. A peripheral blood sample was sent for routine evaluation. The patient presented a normal complete blood count, with a hemoglobin level of 15 g/dL, leukocytes at 5,790/mm^3 (lymphocytes 2,310/mm^3) and 189,000/mm^3 platelets. In the immunophenotypic analysis of the peripheral blood, 30.8% of T lymphocytes were identified, with a CD4/CD8 ratio of 1.4:1. No T cells with the phenotype typically seen in Adult T-cell Leukemia/Lymphoma (ATLL), which was the reason for the sample being sent, were observed. No significant antigenic losses were found in CD4 T cells, and there was no expression of CD25. The TRBC1/TRBC2 ratio in CD4 T cells was found to be 0.85:1.0. Unexpectedly, a T cell population with moderate CD8 expression and a "Large Granular" T phenotype (CD2++/CD3++/partial CD5/CD7+++, CD8++/CD56+++/heterogeneous CD57/CD16 negative/TCR gamma-delta negative) and monoclonal for TRBC2, corresponding to 5.3% of the total leukocytes in the sample (306 cells/mm^3), was observed. **Discussion:** Small populations of monoclonal CD8 T cells can be observed in healthy individuals or during acute viral infections (such as HIV or viral hepatitis, for example), and oligoclonal patterns of TCR gene rearrangements have also been reported in patients with acute infectious mononucleosis (Epstein-Barr virus infection). With the introduction of studying the expression pattern of the constant region of the β1 chain of the T-cell receptor (TRBC1) within a subset of TCR$\alpha\beta$ T cells, it has become more common in routine laboratory practice to find T-cell clones of uncertain significance (T-CUS), which very often have a phenotype resembling T-cell Large Granular Lymphocytic Leukemia (T-LGLL). More recently, the concurrent study of the TRBC2 receptor, along with TRBC1 and a comprehensive T-cell panel, has enhanced the detection of T-cell clones, whether in healthy individuals or in patients with T-cell lymphoproliferative disorders, similar to the routine evaluation of kappa and lambda immunoglobulin light chains for the detection of clonal B cells. **Conclusions:** The diagnosis of T-cell malignancies is often challenging due to overlapping characteristics with reactive T cells and the limitations of currently available T-cell clonality assays. In this case, we suggest ongoing immunophenotypic monitoring, as the patient is asymptomatic, and we emphasize the important point that T-cell clonality by itself, in isolation, is not necessarily indicative of malignancy.

https://doi.org/10.1016/j.htct.2025.104004

14

LLA-B COM FENÓTIPO SUGESTIVO DE REARRANJO DO GENE DUX-4

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Relato de caso: A classificação de neoplasias hematológicas da Organização Mundial de Saúde (OMS) de 2022 incorpora em um grupo, novos subtipos de anormalidades genéticas presentes nas Leucemias Linfoides Agudas B (LLAs-B), incluindo os rearranjos dos genes DUX4, MEF2D, ZNF384 e NUTM1. Os rearranjos envolvendo o gene DUX4 são as lesões genéticas mais frequentes deste grupo e apresentam de forma mais característica correlação com um imunofenótipo específico. Sua identificação é fundamental para a estratificação de risco e, em alguns casos, para otimização da estratégia terapêutica. As LLAs-B com rearranjo envolvendo o gene DUX4 compreendem cerca de 20% das LLAs-B com cariótipo normal. São proporcionalmente mais prevalentes em adolescentes e adultos jovens. Estão associadas a um bom prognóstico mesmo na presença de outros fatores de risco adverso como deleção associada do gene IKAROS e pesquisa de Doença Residual Mensurável (DRM) positiva no início da terapia. É uma alteração genética que pode estar associada à