

time of diagnosis. FISH analysis for IGH/FGFR3 t(4;14), IGH/MAF t(14;16), and p53 gene deletions (17p13.1) showed no abnormalities. Based on international criteria for plasma cell leukemia (PCL)—defined as $\geq 20\%$ and $\geq 2000/\mu\text{L}$ plasma cells in peripheral blood—the diagnosis of primary PCL was confirmed. The patient's clinical presentation, including hypercalcemia, anemia, and osteolytic lesions, supported the diagnosis. **Discussion:** PCL is a rare and aggressive form of multiple myeloma characterized by circulating plasma cells. Morphological variability of plasma cells can complicate diagnosis; cells may mimic lymphocytes, monocytes, or hairy cells. Key diagnostic tools include bone marrow studies, flow cytometry, and a comprehensive myeloma profile, including serum protein electrophoresis, immunofixation, free light chain assay, and 24-hour urine analysis. **Management and Conclusion:** Given the poor prognosis and aggressive nature of PCL, treatment was initiated promptly with a bortezomib-based regimen. PCL highlights the importance of accurate diagnostic methods to guide timely and appropriate therapy.

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COEXISTENCE OF CHRONIC MYELOMONOCYTIC LEUKEMIA AND LYMPHOPROLIFERATIVE DISORDER: THE DIAGNOSTIC ROLE OF IMMUNOPHENOTYPING IN PERIPHERAL BLOOD

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Introduction: The association of chronic myelomonocytic leukemia, with bone marrow infiltration by chronic lymphoproliferative diseases has been reported in the literature mainly by case reports, and documented through bone marrow biopsies. More recently, as the diagnosis of CMML can be made by peripheral blood immunophenotyping, this association could be detected more frequently. This association has been included in the ICC classification of subtypes of CMML.¹ **Case Description:** Male patient, 72 years old, was hospitalized due to pneumonia. At that time, the peripheral blood (PB) count showed leukocytosis with monocytosis and thrombocytopenia, which did not resolve after the pneumonia was cured. Reviewing former PB counts, monocyte counts $>10\%$ could be detected. In 2024, no lymphadenopathy could be observed, but the spleen was 2 cm below the costal margin. **Tests during most recent hospitalization:** Creatinine: 0.9 mg/dL, Ferritin: 46 ng/mL, Serum iron: 67.7 $\mu\text{g/dL}$, ALT: 34 U/L, Vitamin B12: 794 pg/mL, Folic acid: 6.01 ng/mL, Beta-2 microglobulin: 2244 ng/mL (normal <2164), LDH: 320 U/L (normal), Protein electrophoresis: monoclonal peak of 1.46 g/dL; Serum immunofixation: IgM/Kappa. **Bone marrow aspirate:** Hemodiluted.

M:E ratio: 4.28. Lymphocytes: 15%. **Bone marrow biopsy:** markedly hypercellular marrow tissue, granulocytic hyperplasia with maturation arrest. Scattered CD34+ cells. Interstitial infiltration by low-grade lymphoma (lymphocytes and plasma cells) with IgM Kappa on immunohistochemistry. Likely Waldenström macroglobulinemia, with marginal zone lymphoma as differential diagnosis. Immunophenotyping suggested. **Bone marrow immunophenotyping (October 2024):** Monocytosis of 9.1% composed of mature elements. Classical monocytes: 86.1%, Intermediate: 10.8%, Non-classical: 3.1%. Polyclonal plasma cells: 0.15%. Monoclonal B lymphocytes with the following phenotype: CD45+++, CD19++, CD20++, CD43+, CD79b++, CD200++, partial IgM (41%), and predominance of kappa light chain. **Peripheral blood immunophenotyping (November 2024):** Monocytosis of 22.3% composed of mature elements. Classical monocytes: 97.2%, Intermediate: 2.0%, Non-classical: 0.4%. 3.2% monoclonal B lymphocytes with phenotype: CD45+++, CD19++, CD20+++, CD79b++, CD200++, IgM+++, partial CD23 (30%), kappa light chain. **Comments:** In the last years, new strategies for the study of subtypes of monocytes using flow cytometry have been developed. This permitted to better understand the role of these cells in non-neoplastic inflammatory diseases as well as the detection of neoplastic clones. In the present case, the diagnostic work-up in a case with CMML using PB immunophenotyping by multiparametric flow cytometry permitted also the detection of a small lymphoid clone of Waldenström macroglobulinemia. The examination of bone marrow biopsy permitted to diagnose an extensive infiltration of this lymphoproliferative disorder. The patient was treated for macroglobulinemia, and is well and in observation. **Comments:** In this case, peripheral monocytosis detected during infections was known since 2015 but had never been thoroughly investigated. Only during the last infectious episode was the patient referred to a hematologist. After bone marrow immunophenotyping—of which the sample was very diluted—CMML was suspected, and peripheral blood testing confirmed the diagnosis. The monoclonal peak was an incidental finding, but immunophenotyping allowed the diagnosis of chronic lymphoproliferative disease. However, the true extent of this disease in the bone marrow was only assessable through biopsy. The co-existence of myeloproliferative neoplasms or CMML with chronic lymphoproliferative diseases is a rare event (1–2% of chronic myeloproliferative neoplasms). According to limited literature, these appear to be independent neoplasms, not derived from the same stem cell. Immunophenotyping (blood or bone marrow) is the most sensitive technique for detection.

Table 1 The evolution of PB counts.

	11/12/2025	04/16/2019	08/22/2021	01/05/2022	05/02/2024	08/2024	09/2024
RBC	3,81	3,82	3,18	3,89	3,73	3,63	3,7
HB	12,7	12,1	10,2	12,2	12	11,5	12,2
HCT	36,9	37,9	30,70	38,4	36	35,1	36,7
MCV	96,9	99,21	96,4	98,7	96,5	96,6	99,5
LEOCOCYTE	4200	6680	13940	7250	15660	26200	8100
BAND CELLS	42	67	0	73	626	1048	
SEGMENTED	1722	3073	10804	2826	9396	20436	4540
EOSINOPHILS	42	67	0	73	0	0	
BASOPHILS	0	0	42	73	0	0	
TYPICAL LYMPHOCYTES	1680	1536	1450	1740	3289	2620	14600
ATYPICAL LYMPHOCYTES	0	935	0	0	0	0	
MONOCYTES	714 (17%)	1002(15%)	1645 (11,8%)	2538 (35%)	2349 (15%)	2096 (8%)	1780
PLATELETS	140.000	150.000	88.450	121.000	53.280	59.140	101.000

References:

1. Valent P, Orazi A, Savona MR, Patnaik MM, Onida F, van de Loosdrecht AA, et al. Proposed diagnostic criteria for classical chronic myelomonocytic leukemia (CMML), CMML variants and pre-CMML conditions. *Haematologica*. 2019;104:1935-49.
2. Hauck G, et al. Clinical relevance of chronic myelomonocytic leukemia subtypes: experience from a single institution. *Acta Haematol*. 2013;129:187.
3. Torregossa JM, et al. Chronic myelomonocytic leukemia: an underdiagnosed disease. *Med Clin (Barc)*. 2015;145:317.
4. Rybski KJ, et al. Histopathologic spectrum of chronic myelomonocytic leukemia in the modern era. *Int J Surg Pathol*. 2023;31:415.

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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/mm³ (lymphocytes 2,310/mm³) and 189,000/mm³ 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/mm³), 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 β 1 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.

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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 Linfóides 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