Myeloid / Lymphoid Neoplasm with PDGFRA Rearrangement Presenting as T-LBL

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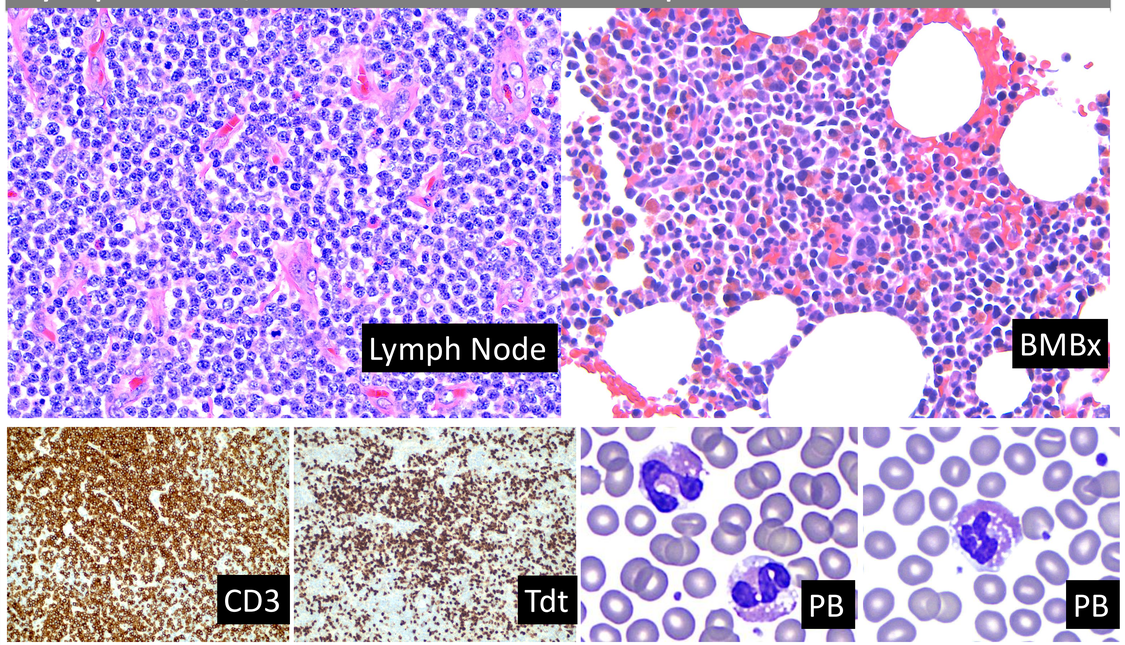
Introduction

- Myeloid/lymphoid neoplasms with eosinophilia (M/LNs-Eo) and rearrangements of PDGFRA, PDGFRB, or FGFR1, or with PCM1-JAK2 genetic variants constitute a rare but well-defined category of hematologic malignancies recognized by the WHO classification.
- Myeloid/lymphoid neoplasms with PDGFRA rearrangements are the most frequent of these disorders and is most common in middle aged males. These neoplasms are extremely rare in the pediatric population.
- The majority of cases are characterized by cytogenetically cryptic deletion of 4q12, resulting in formation of the FIP1L1::PDGFRA fusion gene, although other partner genes have been described.

Case Presentation

- 72 year old male with history of DLBCL diagnosed in Sept. 2016 and treated with R-CHOP + radiotherapy. In remission since March 2017.
- May 2021 follow-up showed enlarged submandibular lymph node without B symptoms.

Lymph Node, Bone Marrow and Peripheral Smear



Right submandibular LN excisional Bx "T-LBL" — May 7th 2021

May 19th, 2021 — **BMBx – Negative for leukemia/lymphoma. No PB eosinophilia. Normal karyotype.**

Started chemotherapy with Hyper CVAD (8 cycles). — May 31st 2021

December 20th 2021 — **Finished chemotherapy.**

Started maintenance therapy with POMP. — January 28th 2022

December 19th 2023 — **Finished maintenance therapy.**

Left neck LN needle Bx. "Recurrent/persistent T-LBL" — August 13th 2024

August 19th 2024 — **BMBx: M/LNs-Eo with FIP1L1::PDGFRA rearrangement.**

Left neck LN excisional Bx: T-LBL with PDGFRA rearrangement, consistent with blast phase of M/LNs-Eo with PDGFRA rearrangement. — September 6th 2024

September 19th 2024 — **Started chemotherapy with miniCVD + TKI.**

CD4 CAR-T cell therapy. — December 10th 2024

Discussion

- We present a case of M/LNs-Eo with PDGFRA rearrangement, which first presented as T-LBL with no marrow involvement.
- The original diagnosis of T-LBL was made because at that time there was no PB eosinophilia and molecular testing was not performed on the samples.
- This demonstrates that M/LNs-Eo can present solely as T-LBL with seemingly normal marrow and PB findings.
- When presented with eosinophilia, clonal processes to consider are CML, CMML, CHES, MDS, TCL, CHL, systemic mastocytosis, and M/LNs-Eo.
- While clonal eosinophilia in association with T-NHL is rare, it highlights the need to expand the differential diagnosis and consider these other potential underlying clonal conditions.
- NGS on our case was done on the relapse BMBx as part of a broader screen for myeloid neoplasms and ultimately revealed the fusion.
- This case highlights the importance of using comprehensive molecular testing panels for an early and accurate diagnosis, but also emphasizes the importance of thoroughly investigating eosinophilia, even in a post-therapy setting, to ensure the correct diagnosis is made.

References

1. Sanam Loghavi, Mrinal Patnaik, Eric Padron, et al. Myeloid/lymphoid neoplasm with PDGFRA rearrangement In: WHO Classification of Tumours Editorial Board. Haematolymphoid tumours [Internet]. Lyon (France): International Agency for Research on Cancer; 2024 [cited 2025 01 05]. (WHO classification of tumours series, 5th ed.; vol. 11). Available from: https://tumourclassification.iarc.who.int/chapters/63.
2. Pozdnyakova O, Orazi A, Kelemen K, King R, Reichard KK, Craig FE, Quintanilla-Martinez L, Rimsza L, George TI, Horny HP, Wang SA. Myeloid/Lymphoid Neoplasms Associated With Eosinophilia and Rearrangements of PDGFRA, PDGFRB, or FGFR1 or With PCM1-JAK2. Am J Clin Pathol. 2021 Feb 4;155(2):160-178. doi: 10.1093/ajcp/aqaa208. PMID: 33367495.