Received: 22 April 2019 | Revised: 16 May 2019 | Accepted: 2 July 2019

DOI: 10.1002/ccr3.2336

CLINICAL IMAGE

Clinical Case Reports Open Access WILEY

Gaucher disease type 1 first recognized in an elderly patient with thrombocytopenia and lung adenocarcinoma

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Abstract

Recognizing Gaucher disease in elderly patients can be challenging. We present a Gaucher disease type 1 case diagnosed in an elderly patient with thrombocytopenia and lung adenocarcinoma. The diagnosis of Gaucher disease was delayed due to lack of familiarity about Gaucher Disease type 1 which can manifest in adulthood.

KEYWORDS

gaucher, lung adenocarcinoma, thrombocytopenia, β-glucosidase

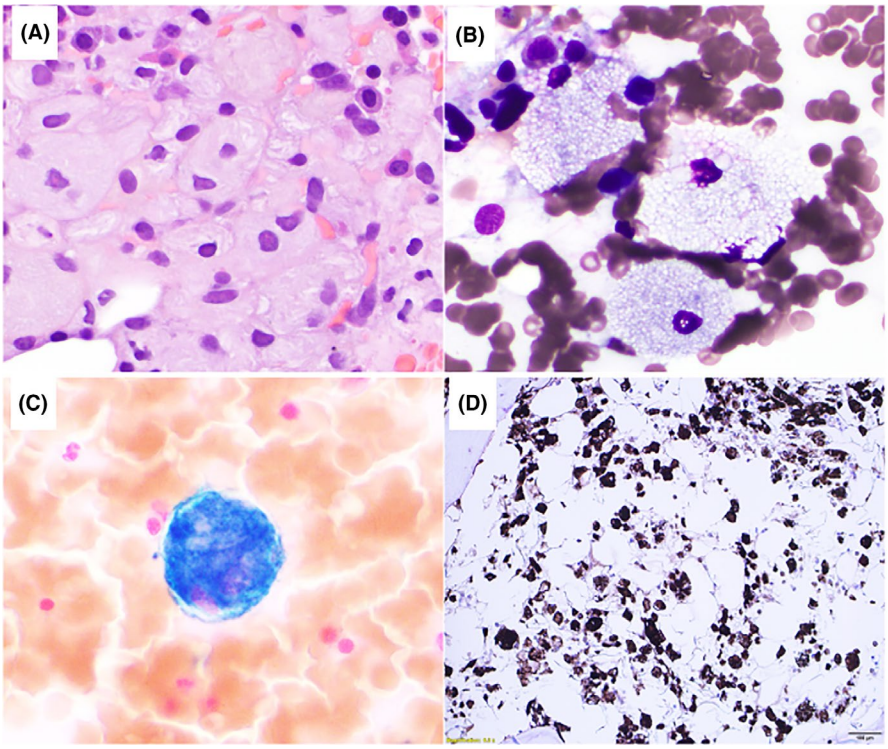

FIGURE 1 A, Bone marrow clot, hematoxylin and eosin stain. B, Bone marrow aspirate, Wright-Giemsa stain. C, Bone marrow aspirate, Prussian blue iron stain. D, Bone marrow core biopsy, CD68 KP1 antibody

A 71-year-old man with a 6-year history of thrombocytopenia presented with epistaxis. Examinations showed a low platelet count and lesions in lung, pleura, and left ethmoid sinus. The liver and the spleen were unremarkable. Biopsies revealed lung adenocarcinoma with metastasis to the left ethmoid sinus. Bone marrow biopsy and aspiration demonstrated histiocytes with wrinkled paper-like fibrillar cytoplasm, occupying 25% of the marrow cellularity (Figure 1A, 1B). These cells were diffusely positive for Prussian blue iron stain (Figure 1C). Immunohistochemistry showed these cells were positive for CD68 (Figure 1D), and were negative for cytokeratin 7. β-glucosidase level in leukocytes was 0.9 nmol/h/mg (normal ≥ 8.7 nmol/h/mg), diagnostic for Gaucher disease type 1.

Gaucher disease is a rare disease. There is a lack of familiarity about Gaucher disease type 1 which can manifest in adulthood.[1] The absence of hepatosplenomegaly and family history in this patient made it less likely that Gaucher disease be considered.[1] However, the histiocytes had hallmarks of Gaucher cells, and showed diffuse iron staining,[2] in contrast to mimickers such as carcinoma cells or histiocytes from normal bone marrow and other lysosomal storage diseases. When the characteristic features are observed, further workup for Gaucher disease should be performed.

CONFLICTS OF INTEREST

None declared.

AUTHOR CONTRIBUTIONS

Dr Wen Shuai: main writer of the manuscript. Dr Celeste Estefania Wagner and Dr Narittee Sukswai: involved in first round of writing the manuscript. Dr L. Jeffrey Medeiros and Dr Carlos Bueso-Ramos: edited the manuscript. Dr Thein Hlaing Oo: supervision of patient care and manuscript writing.

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How to cite this article: Shuai W, Wagner CE, Sukswai N, Medeiros LJ, Bueso-Ramos C, Oo TH. Gaucher disease type 1 first recognized in an elderly patient with thrombocytopenia and lung adenocarcinoma. *Clin Case Rep*. 2019;7:1804–1805. https://doi.org/10.1002/ccr3.2336