

Images in Allergy

Delayed Diagnosis of Good Syndrome

Jessica D. Gereige, MD, Paul J. Maglione, MD, PhD, and Miren Guenechea-Sola, MD *Boston, Mass*

An 82-year-old man presented with worsening vision in his left eye for 2 weeks. He was found to have cytomegalovirus (CMV) retinitis and was treated with intravenous valganciclovir. He has a history of blindness in his right eye thought to be due to an undiagnosed infectious retinitis in the past. His medical history is otherwise notable for a “mass in his chest,” which was incidentally found on imaging more than a decade prior when he was treated for pneumonia. Records obtained from the outside hospital showed biopsy results consistent with thymoma type AB, a noninvasive tumor with low risk of metastasis. A chest x-ray revealed an anterior mediastinal mass, as shown in Figure 1.

Because CMV retinitis is an opportunistic infection that occurs mainly in immunocompromised patients, an immunologic

(1250; 57%; 180-865; 15% to 36%) with an inverted CD4:CD8 ratio of 0.3 (ref 0.9-3.0), and CD19+ B-cell count of zero (ref 25-285; 1% to 21%). He had normal CD16+CD56+ natural killer cells. Immunoglobulin levels were uniformly low: IgG 183 mg/dL (ref 700-1600 mg/dL), IgA 54 mg/dL (ref 70-400 mg/dL), IgM 13 mg/dL (ref 46-304 mg/dL).

The patient was diagnosed with Good syndrome (GS) and started on immunoglobulin replacement, prophylactic antibiotics, and after completion of his CMV retinitis treatment, antiviral prophylaxis.

GS is a rare syndrome defined by the presence of thymoma and immunodeficiency. It was first described in 1955 by Robert Good who played a crucial role in identifying the thymus' role in the immune system.

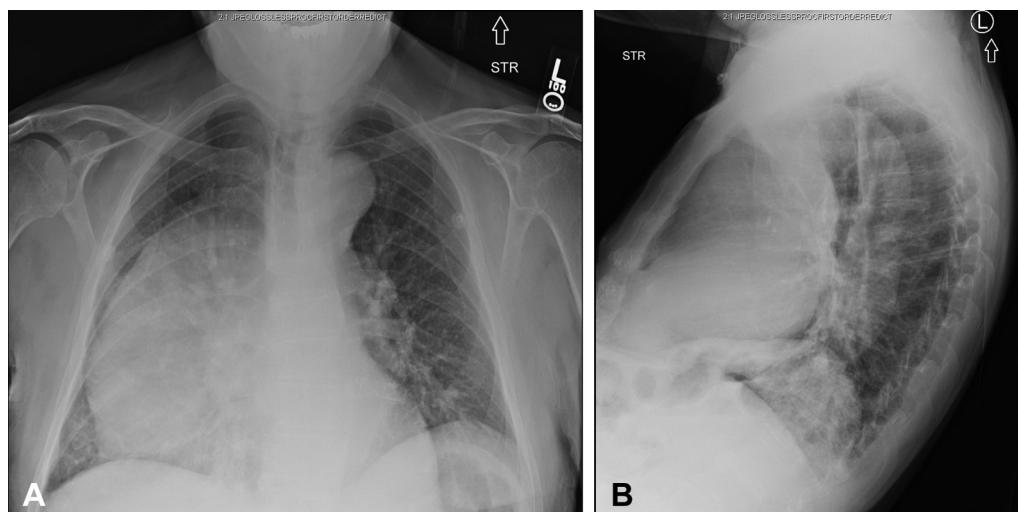


FIGURE 1. **A:** Posterior-anterior (PA) and, **B:** lateral chest x-ray. Chest mass measuring 15.7 × 16.2 cm. The ability to visualize the hilar vessels through the mass (known as the hilum overlay sign) on the PA view, and the obliteration of the retrosternal space and the preservation of the posterior mediastinal lines on the lateral view are radiographic signs consistent with an anterior mediastinal mass.

workup was initiated. An HIV test was negative. Lymphocyte subsets revealed normal CD3+ T cells (1754; 84%; ref 760-2130; 58% to 85%), a reduction of CD4+ T-cell subsets (373; 17%; ref 430-1185; 31% to 62%), an elevation of CD8+ T cells

Patients with GS usually present after the fourth decade of life. Presenting symptoms may be related to the thymoma, either due to its mechanical effects such as cough, dysphagia, and superior vena cava syndrome, or secondary entities such as

Department of Pulmonary, Allergy, Sleep & Critical Care Medicine, Boston Medical Center, Boston University School of Medicine, Boston, Mass

No funding was received for this work.

Conflicts of interest: The authors declare that they have no relevant conflicts of interest.

Received for publication October 10, 2019; revised manuscript received and accepted for publication November 1, 2019.

Available online ■ ■

Corresponding author: Jessica D. Gereige, MD, Boston University School of Medicine, 72 E. Concord St., R 304, Boston, MA 02118. E-mail: jessikag@bu.edu.

J Allergy Clin Immunol Pract 2019;■:■-■.

2213-2198

© 2019 American Academy of Allergy, Asthma & Immunology

<https://doi.org/10.1016/j.jaip.2019.11.019>

myasthenia gravis and pure red cell aplasia. Patients have an antibody deficiency and differ from patients with common variable immunodeficiency (CVID) in that they have very few or absent B cells. GS is also characterized by CD4 lymphopenia, inversion of the CD4:CD8 ratio, and reduced T-cell mitogen proliferation.

Similar to patients with CVID, patients with GS may present with recurrent sinopulmonary infections. However, their CD4 lymphopenia also leads to opportunistic infections normally not seen in patients with CVID. Notably, opportunistic infections in GS differ from patients with HIV in several ways: (1) they occur at significantly higher CD4 counts compared with HIV (CMV retinitis is documented in patients with HIV with CD4 counts <50, but occurs in patients with GS at CD4 counts >300), (2) the most common opportunistic infections include CMV retinitis and colitis, and mucocutaneous candidiasis, and (3) there have been no documented cases of cryptococcus or systemic

fungal infections, and toxoplasmosis and disseminated tuberculosis are rare in patients with GS.

Thymectomy is recommended for patients in whom a margin-negative resection is feasible. However, the immunologic abnormalities leading to immunodeficiency do not correct after thymectomy, and we recommend an immunological evaluation for all patients found to have a thymoma. Treatment of the immunodeficiency consists of immunoglobulin replacement, targeted antimicrobial prophylaxis, and appropriate vaccination with avoidance of live vaccines.^{1,2}

REFERENCES

1. Malphettes M, Gérard L, Galicier L, Boutboul D, Asli B, Szalat R, et al. Good syndrome: an adult-onset immunodeficiency remarkable for its high incidence of invasive infections and autoimmune complications. *Clin Infect Dis* 2015;61:e13-9.
2. Multani A, Gomez CA, Montoya JG. Prevention of infectious diseases in patients with Good syndrome. *Curr Opin Infect Dis* 2018;31:267-77.