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Clinical Presentation and Successful Treatment of a Patient with Anti-SUMO-activating Enzyme Subunit 1 (SAE1) Dermatomyositis

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Abstract Anti-SAE1 antibody is a rarely reported, myositis-specific antibody, which is most clinically associated with affecting the gastrointestinal tract, skin, and lungs. This case demonstrates a patient presenting to the hospital with signs of dermatomyositis, later found to be anti-SAE1 positive. He was promptly treated with immunosuppressive medications, including IVIG, a novel therapeutic agent for dermatomyositis. This case further elucidates the importance of identifying anti-SAE1-positive patients in dermatomyositis.

Keywords: *dermatomyositis, anti-SAE1, polymyositis, antibody, immunology*

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1. Introduction

Dermatomyositis (DM) is a rare, idiopathic autoimmune disease with a largely elusive pathology that is usually characterized by proximal muscle weakness, distinctive rashes, and extra muscular manifestations [1]. Several myositis-specific antibodies have been identified, including anti-Mi-2, anti-TIF1-γ, anti-NXP-2, anti-MDA-5, and anti-SAE (SUMO-Activating-Enzyme). Each antibody has value as some are associated with certain clinical manifestations of DM. This case demonstrates a rare myositis-specific antibody that is seldom reported, anti-SAE1. This patient had several organ-specific manifestations related to this antibody subgroup that are not commonly seen together, which include pulmonary, GI, musculoskeletal, and cutaneous involvement [2].

2. Hospital Course

A 27-year-old male with no significant past medical history presented to the emergency department with a 3-month history of bilateral upper and lower extremity weakness, dysphagia, shortness of breath, and generalized fatigue. Physical exam was notable for erythematous, swollen upper eyelids, scattered hypopigmented lesions in the upper arms, chest, and dorsal hands, and decreased strength in the upper and lower extremities. Initial labs on presentation were significant for a white blood cell count of 12 (thousand/uL), hemoglobin of 10 g/dL, and elevated ESR and CRP at 55 mm/hr and 38 mg/dL, respectively. CT chest depicted peripheral infiltration in the lower lungs, concerning for pneumonia or an inflammatory process (Figure 1). He was promptly started on IV ampicillin/sulbactam and admitted to the hospital.

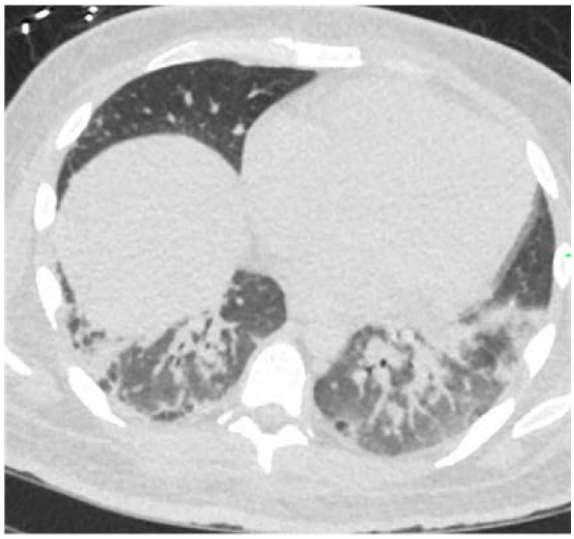

Figure 1. Ground glass opacities with subpleural thickening, no peripheral honey-combing seen

He was previously being evaluated for possible dermatomyositis at an outside, out-of-state facility. At that hospital, a biopsy was performed, which showed interface dermatitis. A myositis panel was obtained on discharge with results pending, and he was given a prednisone taper. He was unfortunately lost to follow-up. During this admission, an autoimmune workup demonstrated an ANA of 1:640, a slightly positive rheumatoid factor at 41

IU/mL (normal <15 IU/mL), a positive SSA 52 IgG antibody of 355 AU/mL (normal < 40 AU/mL), and a highly positive SAE1 antibody. Negative autoantibodies included anti-dsDNA, anti-JO1, anti-CCP, NXP-2, MDA 5, and anti-smith.

The patient underwent further evaluation of the pulmonary, GI, and musculoskeletal systems due to concerns for systemic involvement. MRI of the thighs depicted abnormal fluid intensity throughout all the visualized musculature concerning diffuse myositis (Figure 2). The pulmonary team determined that the patient had early-stage interstitial lung disease (ILD), and pulmonary function testing demonstrated a mild restrictive pattern. An EGD revealed erosions scattered throughout the esophagus with intermittent deep ulcerations. The biopsies of these lesions were negative for infection or malignancy. Due to ongoing weakness, the patient started on mycophenolate 1500 mg twice daily, IVIG 2 mg/kg, and prednisone 20 mg. The patient's symptoms improved, and he was discharged in stable condition with close rheumatology follow-up.

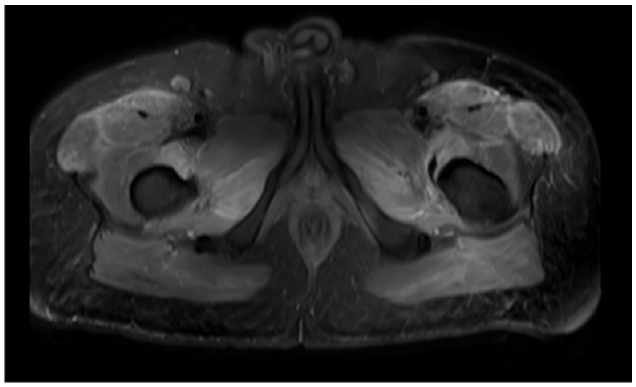

Figure 2. Abnormal fluid signal intensity throughout the musculature of bilateral upper thighs and pelvis

3. Discussion

Anti-SAE1 autoantibody is a rare myositis-specific antibody, with prevalence ranging between 1-8%. In one large European study examining myositis-specific antibodies, only 2% of 1167 patients had anti-SAE1 positive autoantibody. SAE1 antibody targets auto-antigen small ubiquitin-like modifier activating enzyme [3]. Anti-SAE1 is most strongly associated with cutaneous disease (100 % involvement), with the most common presentation being erythema and periorbital edema around the eyelids. Other presenting symptoms are dysphagia, and occasionally, patients will have late-onset muscle weakness and interstitial lung disease. Few cases of anti-SAE1 have reported multi-organ systemic involvement like our patient. In this case, he presented with a heliotrope rash as well as periorbital swelling. He also had erythematous, dry scaling lesions in his arms and trunk. The biopsy of the trunk confirmed interface dermatitis that can be seen in DM. Our patient also had two other manifestations seen in some patients with anti-SAE1 antibodies: interstitial lung disease and dysphagia, both seen in up to 50% of presentations [4]. Interestingly, his serum was anti-JO1 negative, a known antibody that is associated with ILD [5].

IVIG is a treatment already used in DM, especially in refractory cases. However, recently, the FDA approved IVIG therapy after a phase III trial after muscle improvement was seen in the experimental group [1]. This patient had improvement in muscle weakness after two treatments with IVIG. Therefore, this case also illustrates the importance of this treatment in DM.

4. Conclusion

This case is another presentation of a rare autoimmune disease with a unique, seldomly positive autoantibody. The patient suffered from muscle weakness and significant dysphagia for months without proper treatment. He improved promptly with initiating immunosuppressive therapy and steroids, which is expected and consistent with other reports [3]. Providers must identify this subset of SAE1-positive patients and treat this cohort appropriately.

Statement of Competing Interests:

The authors have no competing interests

References

[1] Aggarwal R., Clodi E, Beckmann I., et al. (2023) Trial of intravenous immune globulin in dermatomyositis. *New England Journal of Medicine.*

[2] Betteridge Z., Tansley S., Shaddick G., Chinoy H., Cooper RG., New RP., Lilleker JB., Vencovsky J., Chazarain L., Danko K., Nagy-Vincze M., Bodoki L., Dastmalchi M., Ekholm L., Lundberg IE., McHugh N; (2019) UKMyonet contributors. Frequency, mutual exclusivity and clinical associations of myositis autoantibodies in a combined European cohort of idiopathic inflammatory myopathy patients. *J Autoimmun*. 2019.

[3] de Vries M., Schreurs MWJ., Ahsmann EJM., Spee-Dropkova M., Karim F. (2022) A Case of Anti-SAE1 Dermatomyositis. *Case Reports Immunol.*

[4] Juliette Demortier., Mathieu Vautier., Olivier Chosidow., Laure Gallay., Didier Bessis., Alice Berezne., Nadège Cordel., Jean Schmidt., Amar Smail., Pierre Duffau., Marie Jachiet., Edouard Begon., Jeremy Gottlieb., François Chasset., Julie Graveleau., Myriam Marque., Elise Cesbron., Amandine Forestier., Séverine Josse., Nicolas Kluger., Caroline Beauchêne., Yannick Le Corre., Valentine Pagis., Aude Rigolet., Perrine Guillaume-Jugnot., François-Jérôme Authier., Nelly Guilain., Nathalie Streichenberger., Sarah Leonard-Louis., Samia Boussouar., Océane Landon-Cardinal., Olivier Benveniste., Yves Allenbach. (2023) Anti-SAE autoantibody in dermatomyositis: original comparative study and review of the literature, *Rheumatology.*

[5] Zamora, A. C., Hoskote, S. S., Abascal-Bolado, B., White, D., Cox, C. W., Ryu, J. H., & Moua , T. (2016). Clinical features and outcomes of interstitial lung disease in anti-Jo-1 positive antisynthetase syndrome. *Respiratory Medicine.*