A 26-year-old Man With Treatment Resistant Yao Syndrome

Alexa DeMaio[1] (BS), Gabriella Santa Lucia[1] (BS), Harriet Hinen[2] (MD), Dirk Elston[2] (MD)
[1]Medical University of South Carolina College of Medicine, Charleston, S.C.
[2]Department of Dermatology and Dermatologic Surgery at the Medical University of South Carolina, Charleston, S.C.

Abstract

YAO Syndrome (NOD2 Related Autoinflammatory Condition) has limited data in the literature regarding treatment. Our patient is a 25-year-old-male with NOD2 heterozygosity that has failed several therapies including both first line therapies mentioned in literature.[1] He is now receiving canakinumab with sulfasalazine.[2] To the authors knowledge, canakinumab and sulfasalazine together have yet to be tested on patients with Yao syndrome.[2]

Case History

A 25-year-old male presented with a three-year history of recurrent episodes of tender, slightly pruritic, non-migratory, erythematous eruption often accompanied by myalgias, diarrhea, and sometimes low-grade fever. Multiple biopsies of lesion were consistent with urticaria and labs were grossly unremarkable. The patient failed to respond to antihistamines or steroids. Working diagnosis was chronic urticaria until the patient underwent an autoinflammatory genetic panel through a rheumatology workup, which identified NOD2 heterozygosity, suggesting his symptoms were the result of Yao Syndrome (NOD2 Related Autoinflammatory Condition).[3] Since diagnosis, the patient has failed both first line treatments, sulfasalazine and corticosteroid therapy, as well as colchicine and anakinra, and is now receiving canakinumab with sulfasalazine.

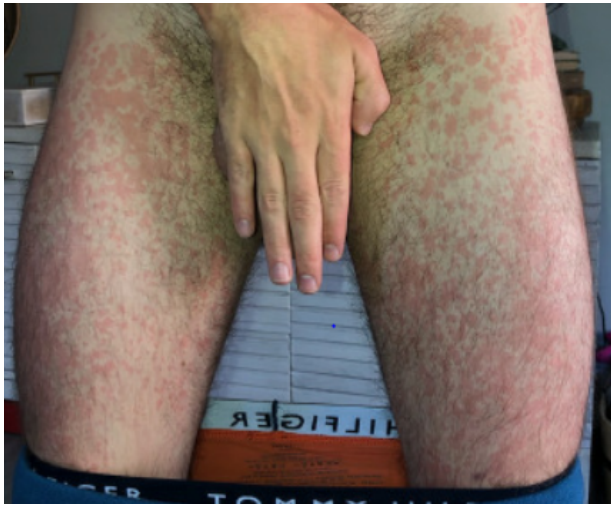

Figure 1: Erythematous urticarial eruption of thighs

References

1. Yao Q, Shen B. A Systematic Analysis of Treatment and Outcomes of NOD2-Associated Autoinflammatory Disease. *Am J Med*. 2017;130(3):365.e13-365.e18. doi:10.1016/j.amjmed.2016.09.028
2. Yao Q. Research letter: Effectiveness of canakinumab for the treatment of Yao syndrome patients [published online ahead of print, 2019 Sep 18]. *J Am Acad Dermatol*. 2019;S0190-9622(19)32757-4. doi:10.1016/j.jaad.2019.09.020
3. Yao Q, Zhou L, Cusumano P, et al. A new category of autoinflammatory disease associated with NOD2 gene mutations. Arthritis Res Ther. 2011;13(5):R148. doi:10.1186/ar3462
4.Yao Q, Shen M, McDonald C, Lacbawan F, Moran R, Shen B. NOD2-associated autoinflammatory disease: a large cohort study. *Rheumatology (Oxford)*. 2015;54(10):1904-1912. doi:10.1093/rheumatology/kev207

Discussion

- Yao syndrome, formerly named NO2-associated autoinflammatory disease.
- Identification across specialties is key to decreasing morbidity, as this disease is believed to have greater prevalence than what has been characterized thus far.[4]
- An auto-inflammatory panel should be considered in patients with periodic urticaria/dermatitis associated with one or more of the following symptoms including fever, polyarthritis, leg swelling, gastrointestinal and sicca-like symptoms.[2]
- Yao is not uncommon, yet effective management strategies are limited due to its recent identification.[3]
- Glucocorticoids or sulfasalazine are first line treatment options with IL-1 and IL-1 antagonist for refractory cases. [1]
- The efficacy of combination canakinumab and sulfasalazine are not noted in literature but are currently managing this patient's disease. Future experimental treatments will be unsupported by literature evidence.