SEVERE COLITIS IN A PATIENT WITH A POLYAUTOIMMUNE SYNDROME - A DIAGNOSTIC AND THERAPEUTIC CHALLENGE

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A 28-year-old man with polyautoimmune syndrome and unclassified inflammatory colitis with recurrent CMV reactivations achieved sustained clinical and biochemical improvement with tofacitinib after multiple treatment failures.

KEY POINTS

- The patient presented with nephrotic syndrome, autoimmune hepatitis/primary biliary cholangitis overlap syndrome, alopecia areata universalis, and developed chronic bloody diarrhea with deep colonic ulcers, fecal calprotectin of 4500 ug/g, and concurrent CMV colitis
- Despite initial improvement with steroids and antivirals, the patient experienced five severe exacerbations with recurrent CMV reactivations and failed multiple therapies including infliximab (loss of response) and azathioprine (adverse reaction)
- After six months of tofacitinib therapy, the patient remains asymptomatic with weight gain, hair recovery, and fecal calprotectin decreased to 1191 ug/g with no new hospital admissions
- Genetic testing revealed a possible pathogenic variant in LRBA gene linked to immunodeficiency type 8 with autoimmunity, with further study ongoing
- The authors conclude this case demonstrates the complexity of polyautoimmune syndrome requiring multidisciplinary management and suggest abatacept may be considered if LRBA deficiency is confirmed

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