

Case Reports

Cureus 2023 Apr 4;15(4):e37142. doi: 10.7759/cureus.37142.

eCollection 2023 Apr.

Cat Eye Syndrome with a Unique Liver and Dermatological Presentation

Maureen Mansur ¹, Thomas J Jacob ², Helen Wong ², Ilya Tarascin ¹

Affiliations

PMID: 37153326 PMID: [PMC10160316](#) DOI: [10.7759/cureus.37142](#)[Free PMC article](#)

Abstract

Cat eye syndrome (CES), also known as Schmid-Fraccaro syndrome, is a complex genetic syndrome with a highly variable phenotype that includes ocular coloboma, anal atresia, preauricular skin tags and pits, heart defects, kidney malformations, dysmorphic facial features, and mild to moderate intellectual disability. We describe a case of a 23-year-old male with a past medical history of CES with short stature, mild learning disability, and some dysmorphic facial features who presented with recurrent pruritus and rashes and had mild liver dysfunction. Furthermore, the patient did not have the classic presentation of CES but a clinically milder expression of the phenotypes. Abnormalities in the abdominal ultrasound prompted an ultrasound-guided liver biopsy, which showed bile ductular proliferation with mild portal inflammation composed of lymphocytes and plasma cells, and bridging fibrosis. The patient's labs showed elevated immunoglobulins with the highest increase observed in IgG, along with negative antinuclear antibodies (ANA), negative anti-mitochondrial antibody, and negative hepatitis A/B/C but a weak positive anti-smooth muscle antibody (ASMA). These findings indicated that the patient most likely had autoimmune hepatitis (AIH) or an overlap syndrome with primary sclerosing cholangitis (PSC). The patient was initially treated with steroids and antihistamines for pruritus, which led to some clinical improvement. After dermatological evaluation, the patient was diagnosed with atopic dermatitis and was recently started on a dupilumab 600 mg loading dose and would continue with biweekly dupilumab 300 mg injections. This dermatological finding may require additional examination and can be a unique presentation in patients with CES. This case illustrates that even patients with milder CES expression can

experience intense dermatological complications if not effectively managed. CES is a multifactorial disease that requires intervention from multiple specialists. Therefore, primary care physicians must be aware of the potential complications of CES and make adequate referrals to closely monitor patients' symptoms.

Keywords: atopic dermatitis; autoimmune hepatitis; cat eye syndrome; chatgpt; livedo racemosa; primary biliary sclerosis; psoriasis; schmid-fraccaro syndrome.

Copyright © 2023, Mansur et al.