

A Distinctive Hepatic and Dermatological Manifestation in Cat Eye Syndrome: A Case Report

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Introduction

- Cat Eye Syndrome can present with variable phenotypes:
 - Classic Triad** = Iris coloboma, Anal atresia and Periarticular skin tag/pit.¹
 - Down slanting palpebral fissure, Short Stature, and mild developmental delays.²
 - Renal and Heart Malformations (Anomalous Pulmonary Venous Return).²
- Dx: Karyotyping, FISH, or aCGH.
- Tx: No cure → Symptom Managements

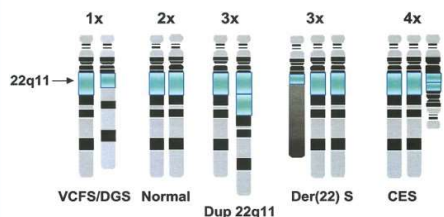


Figure 1: Inverted duplication of the short arm (p) and proximal long arm (q) of chromosome 22 → Three or four times in Chromosome 22.

Patient Presentation

23 y/o M with PMHx of CES with short stature, chronic transaminitis, and recurrent pruritis.

- Review of Systems:** +Abdominal distension, +excessive flatulence, +chronic pruritis, -nausea/vomiting/diarrhea/abdominal pain.
- PE:**
 - General:** Short stature with dysmorphic facial features.
 - Heart:** +S1/S2, RRR, no murmurs.
 - Lungs:** Clear to auscultation, no wheezing.
 - Skin:** Dry skin with generalized scratch marks over his body.
 - Abdomen:** Soft, non-tender distended abdomen with a 1-cm, palpable hepatomegaly.

Disease Course

2013

Cat Eye Syndrome
Diagnosis Established

- Ophtho: b/l myopia.
- Cardiac: Normal EKG.
- Renal: Renal and Bladder ultrasound normal.

2018

Acute Rash Flare

- Multiple plaques on neck, chest, abdomen, back, b/l UE and LE.
- LLE Edema > RLE.
- Diagnosis unclear.

2023

Follow up visits

- Complains of **multiple episodes of pruritis** unresolved with triamcinolone and antihistamines.
- Dermatology referral given → Atopic Dermatitis diagnosed → Biweekly **dupilumab** 300 mg injections → symptoms improving.
- Abdominal distension MiraLAX given.

Elevated LFT Work Up

History:

- No family history of liver disease.
- Denies any alcohol/tobacco/illicit drug use.
- Denies hepatotoxic medication.

Labs:

- Viral Hepatitis A/B/C Panel: Negative
- Elevated Immunoglobulins
- ANA, Anti Mitochondrial Antibody: Negative
- Anti Smooth Muscle Antibody (ASMA): titer 1:20 (weak positive)

Imaging:

- Abdominal Ultrasound:** Increased echogenicity of the portal triads throughout the liver.
- MRCP:** No intrahepatic biliary ductal dilation and no hepatic parenchymal abnormality. Bile duct and common duct morphology not well evaluated due to artifacts due to excessive movement.
- Liver Biopsy:** Bile ductular proliferation with mild portal inflammation and bridging fibrosis. Portal areas with mild inflammation composed of lymphocytes and plasma cells

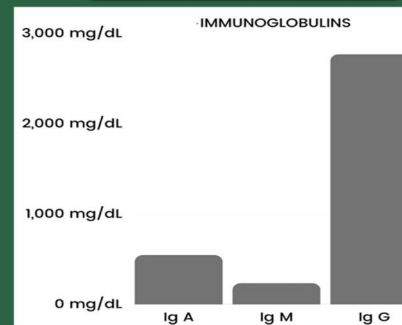


Figure 2: Immunoglobulin Levels (IgA, IgM, IgG)

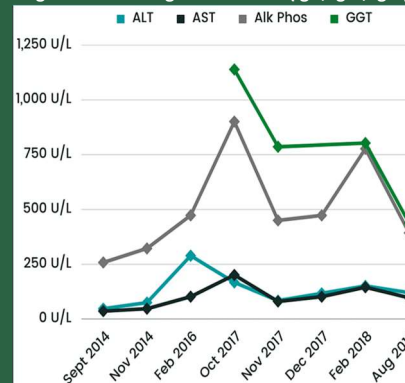


Figure 3: Liver Function Test Trends 2014 -2018

Discussion

- Importance:** Highlights long term problems that may arise in CES patients:
 - Atopic Dermatitis:** 2nd case that associates with CES.³
 - Possible **Autoimmune Hepatitis or AIH-PSC Overlap:** Score of 15 in the Revised Original Scoring System of the International Autoimmune Hepatitis Group.⁴
 - Risk for other Autoimmune Conditions.
- Future Tests:** MRCP with sedation. Repeat ASMA level.
- Tx Optimization:** Corticosteroids + Azathioprine → Relapses and withdrawal to treatment.⁵
 - Adding ursodeoxycholic acid (UDCA)⁵

Take Home Points

- Renal/bladder US, Cardio and Ophthalmologic evaluation on diagnosis.
- Monitor for dermatological problems and LFTs trending earlier.
- Higher suspicion for Autoimmune conditions.

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