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

Disease Course
- **2013** Cat Eye Syndrome
  - Diagnosis Established
  - Ophth: 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.

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.
  - Dx: Karyotyping, FISH, or aCGH.
  - Tx: No cure → Symptom Managements

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

Imaging:
- VCFs/DGS Normal
- Dup 22q11
- Der(22) S CES

History:
- No family history of liver disease.
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Figure 1: Inverted duplication of the short arm (p) and proximal long arm (q) of chromosome 22 → Three or four times in Chromosome 22.

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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

References