CELIAC DISEASE AND MZ ALPHA-1-ANTITRYPSIN DEFICIENCY IN A TODDLER WITH ROTHMUND-THOMSON SYNDROME

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BACKGROUND

- Rothmund-Thomson syndrome (RTS) is characterized by a rash that progresses to poikiloderma: thinning hair; poor growth; skeletal and dental pathologies; cataracts; and an increased risk for cancer, particularly osteosarcoma.
- GI tract involvement: chronic vomiting and diarrhea, sometimes requiring feeding tubes. However, reason for this is unknown and it is considered part of the syndrome.

CASE PRESENTATION

- A 20-month-old female with PMH of Rothmund-Thomson syndrome (RTS) presented to pediatric gastroenterology clinic due to persistently elevated transaminases.

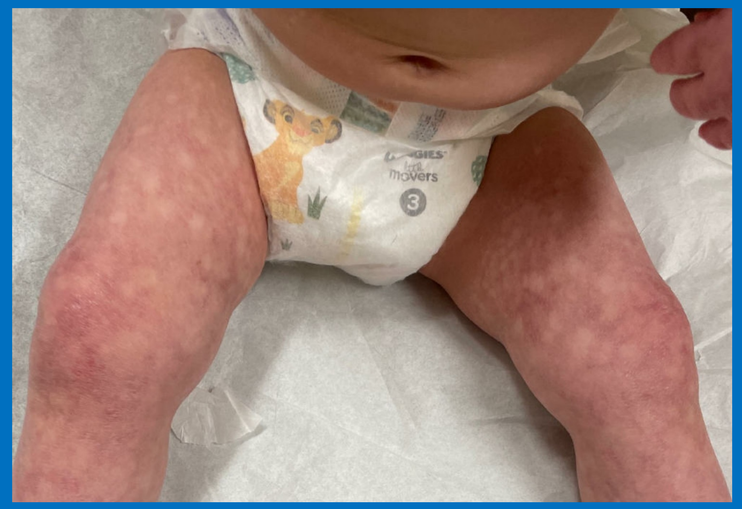

Fig 1: 20-month-old female with RTS and characteristic rash present on the legs.

LAB EVALUATION

- ALT : 101 U/L, AST: 109 U/L
- RUQ abdominal ultrasound: Normal
- Normal Total bilirubin, GGT, Ceruloplasmin, CMV, Hepatitis B and C serology, CK
- Alpha-1-Antitrypsin decreased: 88 mg/dl
- Heterozygous MZ Alpha-1-Antitrypsin phenotype.
- Celiac panel: deamidated Gliadin Peptide IgA 58.5 CU, normal Tissue Transglutaminase IgA and total IgA.
- Upper endoscopy: Duodenal mucosa with mild villous blunting and focal increase in intraepithelial lymphocytes – Marsh 3a.

DISCUSSION:

- There have been rare case reports of Celiac disease but none for Alpha-1-Antitrypsin deficiency association with RTS.
- Since there is known growth delay, chronic diarrhea and vomiting associated with RTS, it may be worthwhile to screen for Celiac Disease and Alpha-1-Antitrypsin deficiency in patients with RTS as treatments can be offered for both and potentially reduce the morbidity associated with the diagnosis of this disease.