Overlapping Clinical Profiles when Juvenile Dermatomyositis Presents with Coeliac Disease

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Introduction

Co-existing immune-mediated conditions including juvenile dermatomyositis (JDM) are well described in coeliac disease. Both conditions can present with overlapping features including dermatological, neuromuscular, nutritional and other manifestations, thereby posing diagnostic challenges necessitating multi-specialty involvement. JDM can present with a variety of non-specific cutaneous manifestations in addition to the pathognomonic Gottron's papules and heliotrope rash. Likewise, coeliac disease may present with a wide variety of non-specific skin rashes, with approximately 10-15% of patients with the classical dermatitis herpetiformis.

Aim

We present a rare case of a three-year old female, with concomitant juvenile dermatomyositis (JDM) and coeliac disease, both conditions diagnosed closely together, presenting with a constellation of overlapping clinical manifestations.

Subjects and Methods

A three-year old girl presented acutely to the Rheumatology clinic with a two-month history of progressive muscle weakness, joint pains, skin rashes and a history of chronic constipation. She was admitted to the paediatric ward for diagnostic workup and management of severe JDM.

Permission for taking of photographs and the consent to share these were obtained from the patients parents.

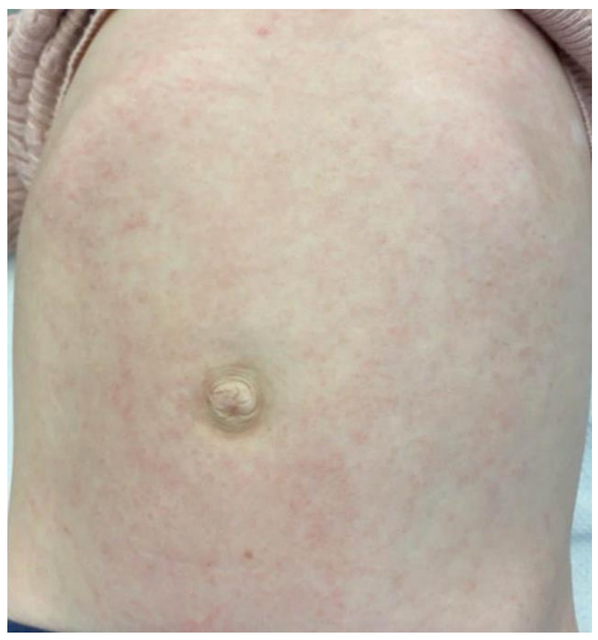

Figure 1 – Maculopapular rash on trunk

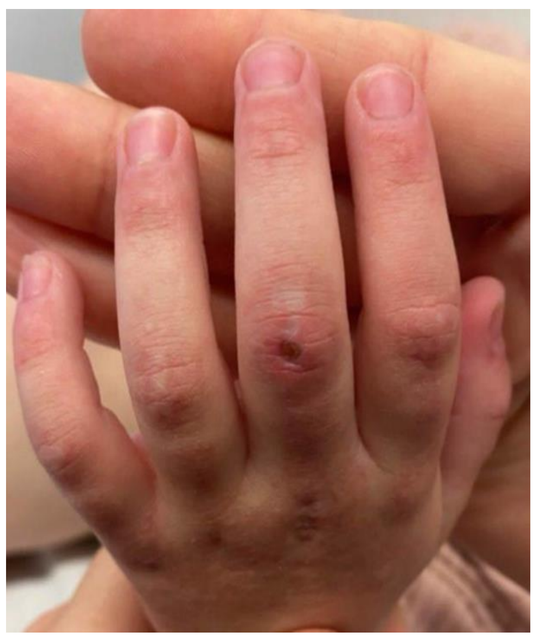

Figure 2 - Gottron's Papules on Left Hand

Results

The patient's symptoms included profound proximal muscle weakness in her neck and upper and lower limbs, a weak voice, unstable gait, arthralgia, myalgia and widespread skin rashes. Two months preceding her illness, she had developed ulcers in her mouth thought to be secondary to hand, foot and mouth disease.

On examination, she had ulcerative skin lesions, periorbital oedema, Gottron's papules on her knuckles and elbows, periungual erythema, livedo on her thighs and a maculopapular rash on her chest. Her muscle enzymes were significantly raised and she had a low creatinine. MRI of both thighs showed moderate-severe inflammation of the muscular, subcutaneous and fascial compartments.

During her admission, she was noted to have significant abdominal distension with distended bowel loops on abdominal imaging. Tissue transglutaminase was elevated (> 250, normal 0-15) with a positive endomysial antibody. MRI of the abdomen did not show evidence of colitis but coincidental findings of a horseshoe kidney and a 18mm lympho-vascular lesion in the liver.

With her complex presentation, a whole exome/targeted sequencing has been considered in addition to myositis specific antibody studies. Alongside her immunosuppressive treatment for JDM, the patient is now making good progress on a gluten-free diet.

Summary and Conclusions

The key points of consideration in this case are:

1) The awareness of co-existing immune-mediated gastrointestinal enteropathies in patients with JDM
2) It is often difficult to map out the contribution of either condition to the overall symptom profile
3) Paediatricians need to be aware that coeliac disease may co-exist with JDM and unless it is looked for early there may be diagnostic delay due to overlapping signs
4) While clusters of autoimmune conditions have been reported in published literature, non-conventional investigations such as whole exome sequencing can be considered for a precise molecular diagnosis which may support clinical decisions and improve patient outcomes
5) With the emergence of robust high throughput technologies, genomic profiling of patients presenting with rare concurrent autoimmune conditions may be useful to identify candidate genes causing overlapping syndromes, inform prognostication and tailored therapeutic approaches in the future.

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