RASH AND WEAKNESS—A CASE OF JUVENILE DERMATOMYOSITIS

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ABSTRACT

Juvenile dermatomyositis (JDM) is a well-known but uncommon paediatric rheumatological condition. It can be missed early in the disease manifestation if not thought of in patients presenting with atypical rash and weakness. We present a case of a 5-year-old girl who developed JDM over a span of 8 weeks before a diagnosis was made. This case demonstrates the nuances of how a paediatric patient's presentation can differ from adults'. A high index of suspicion and early appropriate referral to paediatric rheumatology are paramount for successful management and good prognosis of the condition.

Keywords: Juvenile Dermatomyositis; Gottron's Papules; Heliotrope Rash; Atopic Dermatitis; Bilateral Eye Swelling; Recurrent Falls;

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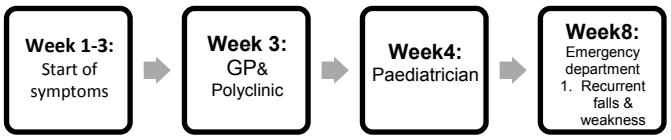
PATIENT'S REVELATION: WHAT HAPPENED?

Figure 1: Progression of events.



Picture 1: Heliotrope rash.

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Picture 2: Vasculitic rash of lower limbs.

Picture 3: Gottron's papules.

Picture 4: Rugged cuticles and periungual erythema.

Z is a 5-year-old Chinese girl who presented with recurrent falls and atypical rash.

First Consultation

Z was first brought by her parents to a general practitioner with complaints of leg rash, bilateral eye swelling, and tiredness for 3 weeks. Her double eyelids were no longer well demarcated due to the swelling. She was less active at home and at school. She did not want to play at the playground, as was her usual routine, and her teacher reported that she seemed quiet and tired in school. She did not complain of pain that limited her play. She was diagnosed with food allergy and advised to go to a polyclinic. Of note, Z had no recent illness or insect bites, nor did she take any new medications or food. There was no low-grade fever, loss of weight, or appetite. She had no significant past medical history and had no personal or familial history of autoimmune disease.

Second Consultation

Z attended the polyclinic the same day. Additional history did not suggest any visual disturbances, red eyes or eye pain. There was no personal or familial history of atopy. Z was afebrile with a normal pulse rate and blood pressure. Eye examination showed bilateral eyelid swelling and equal pupillary light reflex. Dry, erythematous scaly plaques were noted over bilateral shins and knees. Examination of cardiovascular, respiratory, and abdominal systems was unremarkable. Of note, no neurological examination was done. Thyroid and renal panels were performed to evaluate for her tiredness and eye swelling, but returned normal. Her urine dipstick showed trace blood of 1+ but no proteinuria, leucocytes, or nitrite. Her diagnosis was altered to atopic dermatitis and hydrocortisone cream prescribed. A referral letter was made to the paediatric allergy clinic.

Third Consultation

She was seen one week later by a paediatrician from the allergy team. Urine microscopy to investigate the trace haematuria on dipstick was performed, showing no erythrocytes. The rash was the focus of the consult with no mention of the bilateral eye swelling or lethargy. The diagnosis of moderate eczema persisted and Z was prescribed a more potent steroid cream with a 1-month review.

Fourth Consultation

Her concerned parents brought her to the children's emergency department 4 weeks later with the main complaint of recurrent falls and worsening rash. She started falling down while walking and took a long time to climb the stairs at school. She had to use surrounding household items as support to stand. She did not report limitation of exercise due to chest pain or shortness of breath. No choking on food or change in voice was noted. Her rash had progressed despite the use of steroid creams and now involved extensor surfaces of both forearms and knuckles.

On examination, Z walked into the consult room with a waddling gait with no torticollis. Her vital signs were: temperature 36.9°C, heart rate 108/min, blood pressure 120/85mmHg, respiratory rate 20, and pulse oximetry 100% on room air. She had a heliotrope rash over her upper eyelids with periorbital swelling, malar erythema, and a scaly purplish rash over the bilateral extensor surfaces of her limbs. Gottron's papules were noted over bilateral elbows and interphalangeal joints, and examination of her fingers revealed dilated capillary nail beds, ragged cuticles with periungual erythema. There was no nail dystrophy. Cardiorespiratory examination was unremarkable. Neurological examination demonstrated a normal tone and reflexes ++ in all 4 limbs with a normal cranial nerve exam. Weakness was predominantly proximal with power 3+/5 proximally and 4+/5 distally in all 4 limbs. There was no axial weakness but Z tested positive for Gower's sign. There was no shawl sign, V-neck sign, calcinosis, joint swelling, or other systemic involvement. She was admitted for further workup with a provisional diagnosis of juvenile dermatomyositis.

Initial blood investigations showed white blood cell count of 7.83 x 10^9/L, with normal haemoglobin of 12.4g/dL, and platelet count of 432 x 10^9/L. Electrolytes, renal function, and urine microscopy were normal. On retrospect, the false-positive blood on urine dipstick was likely secondary to myoglobinuria. Liver panel showed elevated AST: 178 U/L and ALT: 69 U/L and high LDH: 2292 U/L. Elevated creatinine kinase: 2947 U/L and aldolase: 32.3 U/L suggested myositis. An antinuclear antibody (ANA) test was performed to screen for possible mixed connective tissue disorder and returned normal. MRI of her bilateral thighs with contrast showed diffuse bright T2/STIR high signals with post-contrast enhancement. This was highly specific for muscle inflammation and oedema. There was no cardiac arrthymia on electrocardiogram, and a formal speech and swallowing assessment showed no dysphonia or dysphagia.

Z fulfilled the criteria for definite JDM of moderate severity. Traditional diagnostic criteria by Bohan and Peter in 1975[1,2] requires typical skin findings with 3 other criteria to diagnose definite JDM, and 2 criteria for probable JDM. The criteria include characteristic skin rash with proximal muscle weakness, and elevated muscle enzymes, myopathic changes on electromyography or muscle biopsy. The 2006 international consensus survey expanded the above criteria to include non-invasive tests that demonstrate myositis on MRI or nailfold capillaroscopy abnormalities.[3,4]

Z was quickly commenced on intravenous methylprednisolone and methotrexate to induce remission. Subsequent muscle power and skin rash improved partially over 2 weeks and Z was discharged on subcutaneous methotrexate and high-dose oral prednisolone as maintenance therapy.

GAINING INSIGHT: WHAT ARE THE ISSUES?

Several issues arose as a result of this case:

(i) What are the clinical features of JDM and are there red flags

in this case which should alert primary care providers to its diagnosis?

(ii) How can one differentiate a vasculitic rash of JDM from childhood eczema?

(iii) What are the similarities and differences between juvenile and adult dermatomyositis?

(iv) A short word on the natural history and prognosis of JDM.

MANAGEMENT: HOW DO WE APPLY OUR INSIGHTS IN OUR CLINICAL PRACTICE?

Clinical Features of JDM and Red Flags

JDM affects about 4 in 1 million children each year.[5] Z fit the classic epidemiology of JDM with the peak incidence from 5 to 10 years of age. Girls are more affected than boys with a 2- to 5-fold greater rate.[6] Risk factors for JDM include family history of JDM in first-degree relatives.[7, 8] or a family history of autoimmune conditions such as type 1 diabetes mellitus or systemic lupus erythematosus (SLE)[9] which Z did not have.

JDM can present classically with characteristic rash and weakness as in Z's case or as JDM sine myositis with rash but asymptomatic low-grade muscle inflammation only detectable on muscle biopsy but not clinically. JDM can also present with multisystem involvement of the gastrointestinal, respiratory, and cardiac systems, and may be associated with other rheumatic diseases like SLE.

The onset of JDM is insidious, starting with constitutional symptoms of fatigue, loss of appetite and weight, and low-grade fever for weeks to months. Dermatitis presents early in the disease, with the three most typical cutaneous manifestations being a heliotropic discolouration of the upper eyelids, Gottron's papules, and periungual erythema with capillary loop abnormalities.[10] Weakness usually develops later in the disease and is predominantly proximal with complaints related to weakness of the lower limb and proximal girdle being the most common.[11] This differentiates it from peripheral neuropathies that typically present with distal weakness and associated numbness.[12] If allowed to continue untreated, the weakness may become permanent and may involve axial muscles to cause dysphagia and dysphonia.[13] Rarely, patients have vasculopathy involving cardiac, respiratory, and gastrointestinal systems which confers a poorer prognosis.[14] Cardiac involvement with myocarditis may result in heart failure, cardiac arrthymias, and mortality in severe cases.[15] The turning point in Z's case came when she presented with weakness, resulting in recurrent falls, and an atypical rash not responding to steroids which prompted doctors to relook her diagnosis. A combination of rash, weakness, and constitutional symptoms should prompt primary care providers to consider dermatomyositis.

There were a few atypical features that served as red flags in her case. The 3-week duration of Z's complaints on initial presentation should prompt one to consider a subacute or evolving chronic condition rather than an acute one such as food allergy. Secondly, her symptoms of tiredness were reported by her parents as well as her schoolteacher. This should highlight to a physician that the complaint should be taken seriously and not attributed to an overly concerned parent as they were noticed in two different settings. Thirdly, the sudden eruption of a rash in a child with no atopic history and no new triggers is unusual and should not be attributed to "atopic dermatitis" even though that is a very common rash of that age group. Lastly, there were unexplained symptoms of bilateral eye swelling and fatigue. These symptoms were not adequately explored as the main focus was placed on the rash. This highlights the importance of considering a patient's symptoms in its totality instead of focusing on the most prominent symptom alone.

Initial differentials to consider in this case include nephrotic syndrome which was ruled out by the absence of proteinuria, hypertension, or dependent oedema. Food or drug allergies typically present acutely with urticarial rash and patients should not have constitutional symptoms of fatigue. Urticaria would not be restricted to the lower limbs and its morphology clearly different from the scaly, erythematous rash described in Z's case. A patient with atopic dermatitis usually has some personal or familial atopic history, identifiable triggers, a classic eczematous morphology, and good response to topical steroid, which were all absent in Z's case. Hypothyroidism with pre-tibial myxoedema may account for Z's lower limb rash and fatigue but does not account for her bilateral eye swelling. This was excluded with a normal thyroid function test. Lastly, connective tissue disorders like SLE should be considered in the presence of a vasculitic rash, unexplained eye swelling, and constitutional symptoms but the patient lacked other criteria of arthritis, serositis, mucosal involvement, blood disorders, or a positive ANA test.

How does a rash in JDM differ from a rash of atopic dermatitis in children?

Table 1: Differences between Atopic Dermatitis and JDM.

Feature	Atopic dermatitis	Rash in JDM
Family history	- Personal or familial history of atopy: eczema, allergic rhinitis, asthma [16]	- Personal or familial history of autoimmune diseases
Distribution	- Scalp, cheeks, extensor areas <18 months [16] - Neck, elbow, and knee flexures, wrist and ankle distribution in older children	- Eyelids: Heliotrope rash - Cheeks: malar rash which may be photosensitive - Shawl sign, Gottron's papules, and extensor distribution over limbs
Appearance	- Erythematous when in flare, followed by post-inflammatory hypo/hyperpigmentation	- May be more hypertrophic, raised and palpable - Violaceous hue - Scaly or shiny appearance
Exacerbating factors	- Allergens - Heat	- Photosensitive, worse with sunlight exposure[10]
Characteristic skin features	- None	- Periorbital oedema - Gottron's sign especially over interphalangeal joints - Skin ulcerations or calcinosis[10] - Periungual erythema or dilated nail-bed capillary loops
Response to steroid creams	- Usually responds to steroid cream application	- Little or incomplete response

What are the similarities and differences between juvenile and adult dermatomyositis?

Table 2: Referenced from Textbook of Paediatric Rheumatology, Sixth Edition.[1]

Factor	Similarities	Differences	
		JDM	Adult DM
Epidemiology	- Female predilection in both	- Peak age onset 7.6 years[6]	- Peak onset 40-50 years
Clinical manifestation	- Share many clinical features	- Calcinosis, lipodystrophy, gastrointestinal and cutaneous ulcerations more common[19]	- Interstitial lung and myocardial involvement more frequent[18] - Greater degree of muscle weakness in adult DM - Generally more severe disease activity in adult DM
Association to malignancy	- Only adult DM is associated with malignancy	- No association	- 5 to 7 fold compared to general population[20] - Adenocarcinomas of the cervix, lung, ovaries, pancreas, bladder, and stomach account for 70% of malignancies associated with DM[21]
Response to treatment	- Steroids are mainstay of treatment	- Greater magnitude of response to treatment	- Poorer response
Outcomes	- Both improving with increased access to immunosuppressive therapies	- Lower mortality (<3%) - Functional disability (28–41%)	- Higher mortality (10–26%) - Functional disability (35–60%)

A Short Word on the Natural History and Prognosis of JDM

There are three patterns of disease in JDM.[22] Two-thirds follow a chronic persistent course, and one-third a monocyclic course in which there is one disease episode that responds to standard treatment without relapse. A minority (3%) may have a polycyclic course with multiple remissions and relapses and poorer prognosis. Early treatment is crucial to limit JDM to a monocyclic pattern and prevent late-stage ulcerative skin disease, permanent muscle weakness, or life-threatening major organ involvement. Z seems to be responding to her maintenance therapy one month post treatment and hopefully her disease will remain quiescent.

CONCLUSION

Though paediatric rheumatological conditions like JDM are uncommonly encountered in the primary care setting,[23,24] early diagnosis and treatment can lead to control of these often debilitating diseases before irreversible end organ damage is done.[25] This article highlights a few key lessons for the primary physician.

(i) The need to understand the nuances of a paediatric patent's presentation compared to that for an adult population;

(ii) The importance of evaluating patient's symptoms in its totality instead of focusing on the most obvious symptom;

(iii) The need to reconsider one's diagnosis if there are atypical features; and

(iv) The need to be aware of how paediatric rheumatological conditions present for early referral.

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Ethical considerations

Verbal and written consent was obtained from the patient's parents for the purposes of this publication, including the use of photographs.

Conflicts of interest

The author declares that he has no conflict of interests in relation to this article.

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