Juvenile Dermatomyositis: A Case Study

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KEY WORDS
Juvenile dermatomyositis, pediatric, skin rash, muscle weakness

CHIEF COMPLAINT

A mother reports that her daughter has a rash on her face, an itchy rash on her upper arms, hands, and right shin, as well as fecal incontinence and vomiting after large meals.

HISTORY OF PRESENT ILLNESS

A 7-year-old White girl presented with an erythematous rash located bilaterally on her cheeks and extending over the bridge of the nose along with a purplish rash over her eyelids that had been present for approximately 6 to 8 weeks. In addition, the child had a pruritic bumpy rash on her upper arms, elbows, knuckles, and knees and an elongated purplish area on her right shin. The child's mother stated, "The facial rash seems to worsen after she has been in the sun." The mother also reported that the child had several "bowel accidents" a day, that her stool is "liquid," and that she "frequently vomits after eating a large meal and then resumes eating 15 to 30 minutes later."

The rash first developed after swimming for several hours on a summer day. The erythematous facial rash presented more on the right side than the left and bilaterally on her eyelids. Sun exposure made the rash worse. The rash was associated with pruritus. Topical treatment with diphenhydramine and over-the-counter hydrocortisone did not improve the rash. The patient was seen multiple times by a pediatrician, who prescribed topical steroids for eczema. This treatment was ineffective and the rash continued to progress in severity, extending to the elbows, knees, anterior aspect of both legs, and to the lateral and medial malleolus. The rash became more erythematous and scaly with excoriations as a result of itching. A dermatology consultation was arranged.

MEDICAL HISTORY

The child had a history of gastric reflux, eczema, reactive airway disease, and leg-length discrepancy (her left leg is shorter than the right leg). She had no prior surgeries. All immunizations were current. Medications she was currently taking included Singulair, 4 mg daily, and the following medications on an as-needed basis: Claritin, Sudafed, Nasonex nasal spray, Robitussin DM, ibuprofen, topical diphenhydramine cream, and topical hydrocortisone cream. The patient is on target for all developmental milestones.

BIRTH HISTORY

The patient was born at 38.5 weeks gestation via forceps-assisted vaginal delivery. The mother was

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negative for group B streptococcus and had no known intrauterine maternal infections.

SIGNIFICANT FAMILY HISTORY

Significant family history includes a maternal grandmother with hypothyroidism, a maternal uncle with psoriatic arthritis, a maternal cousin with type I diabetes mellitus, and a maternal great uncle with lupus.

PHYSICAL EXAMINATION

The patient is an alert, cooperative young girl in no apparent distress. Her vital signs are as follows: oral temperature, 36.5°C (97.7°F); heart rate, 105 beats per minute; respiratory rate, 22 breaths per minute; and blood pressure, 113/65 mm Hg. Her weight is 22.5 kg (42nd percentile) and her height is 119.3 cm (27th percentile).

A skin examination revealed that the child had nail fold erythema and nail bed telangiectasia; no palate dilation; Gottron's papules on the proximal interphalangeal joints, knees, elbows, and first two metatarsals bilaterally; cuticular overgrowth; and capillary erythema. Moderate erythema and vasodilation was observed on the eyelids (heliotrope rash), and she had a malar rash. Her pupils were equal, round, and reactive to light; the disks were sharp. A bilateral ankle exam revealed limited range of motion. Her hamstrings were tight and she had a leg length discrepancy of 1 cm, with the right leg longer than the left; in addition, her right shoulder was higher than her left shoulder. A neurologic examination was significant for neck flexor strength, which was 3/5; in addition, upper proximal muscle strength was 3/5 and lower muscle strength was 3/5. She had a negative Gower's sign and was able to sit up with her arms out, crossed, and behind her head. The remainder of her physical examination was unremarkable.

CASE STUDY QUESTIONS

1. What differential diagnoses should be considered for this patient?
2. What are some other possible clinical manifestations of juvenile dermatomyositis that this patient does *not* exhibit?
3. What are the criteria for diagnosis of juvenile dermatomyositis?
4. What initial diagnostic tests are you considering?
5. What treatment plan and follow-up should be recommended?

CASE STUDY ANSWERS

1. *What differential diagnoses should be considered for this patient?*

Skin rash differential diagnoses include psoriasis, eczema, and allergies (Feldman, Rider, Reed, & Pachman, 2008). Based on the "classic rash" on the child's hands, the initial dermatology consultant gave the preliminary diagnosis of juvenile dermatomyositis (JDM). A second dermatological opinion was sought by the family. Laboratory tests for creatine kinase and aldolase were ordered, and these levels were elevated. The second dermatologist agreed with the preliminary diagnosis of JDM and referred the patient to a pediatric rheumatologist. Proximal muscle weakness and hoarseness began to manifest, and the child was admitted to the hospital. Several diagnostic tests were performed, including a magnetic resonance imaging (MRI) scan of the lower extremities and a muscle biopsy of the left vastus lateralis; findings were consistent with JDM. She received two pulses of intravenous methylprednisolone and was discharged home with a prescription for oral prednisone, 40 mg every morning (approximately 1.5 mg/kg/day), hydroxychloroquine, 100 mg every morning, and Prevacid, 15 mg daily.

The JDM pruritic skin rash can resemble an allergic skin reaction. However, the JDM rash generally is less responsive to topical steroid creams (when used alone) than are allergic skin rashes. Systemic steroids result in a greater clinical anti-inflammatory response. Topical steroids and new nonsteroidal creams such as tacrolimus (Protopic) or pimecrolimus (Elidel) may be used as an adjunct to treatment (Rider, Pachman, Miller, & Bollar, 2007b).

The differential diagnoses for the gastrointestinal clinical manifestations (i.e., vomiting, diarrhea, and fecal incontinence) should include viral, bacterial, and parasitic infections (Feldman et al., 2008). In this particular case, diarrhea and vomiting may have been caused by gastroenteritis of the gastrointestinal tract, causing sphincter muscle weakness. Inflammatory bowel disease and celiac disease also should be ruled out (Feldman et al., 2008). The differential diagnoses for the muscle weakness include viral and bacterial infections, systemic lupus erythematosus, juvenile arthritis, dystrophies, and myopathies (Feldman et al., 2008).

The JDM pruritic skin rash can resemble an allergic skin reaction.

2. *What are some other possible clinical manifestations of juvenile dermatomyositis that this patient does not exhibit?*

JDM is a rare, potentially life-threatening systemic vasculopathy of unknown origin that appears to be autoimmune in nature. It primarily affects the skin

and muscles but can affect the gastrointestinal tract, lungs, and heart (Lowry & Pilkington, 2009). Although very serious in nature, the prognosis is generally excellent. The use of early, aggressive corticosteroid therapy has improved lifetime mortality rates from approximately 30% 40 years ago to less than 2% today (Feldman et al., 2008). Some children have a monocyclic course (permanent remission after 2 to 3 years of treatment), others have a polycyclic course (remission and exacerbation periods), and some have a chronic continuous course (Feldman et al., 2008).

The incidence of JDM in the United States is approximately 3 per 1 million children per year. The average age at onset is 7 years, and girls are affected twice as often as boys (Feldman et al., 2008). A genetic predisposition exists, but studies suggest an environmental infectious pathogen trigger, especially in children who report a history of gastrointestinal or respiratory infection in the previous 3 months (e.g., Coxsackie B virus or enterovirus) (Pachman et al., 2005). Some studies suggest a link between ultraviolet sun exposure and initiation of symptoms; however, studies supporting this link have been inconclusive (Manlhiot et al., 2008). Sun exposure may, however, aggravate or exacerbate existing symptoms, especially in sun-exposed areas of the body (Lowry & Pilkington, 2009). Using sunscreen daily and wearing photo-protective clothing is strongly recommended (Feldman et al., 2008; Lowry & Pilkington, 2009).

JDM may be difficult to diagnose because of the variation in clinical presentation. Approximately 50% of children present with rash as their initial symptom and 25% present with weakness as their first symptom (Feldman et al., 2008). Other dermatological presentations may include a v-sign rash or "shawl" sign around the neck that may appear only in the sun-exposed area of the neck and may be photosensitive (Lowry & Pilkington, 2009). Erythematous skin changes and Gottron's papules, such as those observed in this case, may appear as skin thickening or look like "alligator skin" over the metacarpal and phalangeal joints, knees, and elbows. The pruritic rash also may involve the limbs and trunk (Pachman et al., 2005). In the total adult dermatomyositis population approximately 10% to 20% of patients present with amyopathic (or skin only) cases that eventually progress to myositis (Lowry & Pilkington, 2009). Livedo reticularis (marbling effect of the skin) as a result of vasculitis may be present (Pilkington & Wedderburn, 2005).

The inflammatory response is variable. Muscle weakness that is mild may be easily overlooked, and inflammation may be detectable only on an MRI scan (Pilkington & Wedderburn, 2005). Because of the chronic inflammatory response, muscle damage may occur, leading to weakness and atrophy. If muscle weakness becomes profound, the child often will compensate by using larger muscle groups to carry out daily activities. Weakness may only become apparent when the child can no longer climb stairs or get into and out of a car (Pachman et al., 2005). Changes in the child's voice quality, coughing, and dysphagia may be clinical manifestations described by the parents. Muscular weakness can include the esophagus, lower esophageal sphincter, and respiratory muscles (Pilkington & Wedderburn, 2005).

Approximately 30% of children with JDM will have dystrophic calcification affecting pressure points, elbows, knees, digits, and buttocks. Calcification may or may not be a presenting symptom. It usually begins 1 to 3 years after the onset of symptoms. Ongoing, untreated inflammation may contribute to its development (Feldman et al., 2008).

Lipodystrophy, a disorder of the adipose tissue in which selective loss of body fat occurs, is a complication of JDM in approximately 10% to 40% of patients, and the presentation may be generalized, partial, and local. Lipodystrophy presence predicts a chronic disease course with concomitant calcinosis, muscle contractures, and atrophy (Lowry & Pilkington, 2009).

3. *What are the criteria for diagnosis of juvenile dermatomyositis?*

The diagnostic criteria detailed by Bohan and Peter (1975) include the characteristic skin rash and three of the following: symmetrical muscle weakness of the upper and lower proximal muscles, increased levels of serum muscle enzymes, and myopathic electromyography or characteristic pathologic changes revealed by a muscle biopsy. This patient met three of the four criteria at time of preliminary diagnosis. She presented with the classic skin rash, proximal muscle weakness, and elevated levels of serum muscle enzymes. An initial electromyography while she was hospitalized was inconclusive. A lower extremity MRI scan detected muscle inflammation in both thighs. A muscle biopsy subsequently was performed on her left vastus lateralis muscle. Findings from the muscle biopsy provided the fourth criteria necessary for a confirmatory diagnosis of JDM.

4. *What initial diagnostic tests are you considering?*

Blood tests to measure specific markers of muscle inflammation should be ordered including creatine kinase, lactate dehydrogenase, aldolase, aspartate aminotransferase, and alanine aminotransferase (Pilkington & Wedderburn, 2005). A white blood cell count with differential and stool cultures would rule out pathogenic agents responsible for vomiting and diarrhea. To rule out Celiac sprue or inflammatory bowel disease, serologic testing or an intestinal biopsy are necessary (Porth & Matfin, 2009).

Electromyography may demonstrate myopathy and denervation. Because muscle inflammation may be patchy, electromyography is not always diagnostic, as was seen with this patient. MRI scans have become more widely used and can be helpful in choosing the correct muscle biopsy site. Muscle biopsy findings that are consistent with JDM are perifascicular atrophy, perivascular inflammatory infiltrates, internal myonuclei,

and necrosis of muscle fibers (Pilkington & Wedderburn, 2005).

Initial diagnostic tests included CK and aldolase, which were elevated at 1422 units/L and 16 units/L, respectively, and helped confirm a preliminary JDM diagnosis. After referral to a pediatric rheumatologist, this child underwent several additional diagnostic tests; the abnormal findings are listed in the Table.

5. *What treatment plan and follow-up should be recommended?*

Early JDM diagnosis and aggressive pharmacologic corticosteroid treatment are key to favorable outcomes (Pilkington & Wedderburn, 2005). Many practitioners may never see a case of JDM in their careers, and because the presenting symptoms may vary, diagnosis can be a challenge. As in this case, it is not uncommon for as many as four to five practitioners to be consulted before a diagnosis is confirmed (Pachman et al., 2005). Furthermore, some signs associated with a poor prognosis may be subtle and therefore missed. The delayed diagnosis often is accompanied by continued and increasing inflammation and hence potential further systemic damage. Active disease that is left untreated (even for as short a time as 3 to 4 months) leads to a less favorable outcome (Pachman et al., 2006). Feldman and colleagues (2008) state that a delay in the use of corticosteroid treatment "is one of the most important predictors of poor outcome and chronic illness course, including a decrease in bone density and chronic skin disease." Administration of subcutaneous methotrexate and pulses of intravenous methylprednisolone upon initiation of corticosteroid therapy may "shorten the course of the illness, reduce calcinosis, reduce the likelihood of a flare later in the course of the illness, and increase the chance of remission" (Wedderburn & Rider, 2009). Early introduction of subcutaneous methotrexate also may reduce corticosteroid toxicity and result in less weight gain, better growth, and reduced cataract formation (Wedderburn & Rider, 2009).

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Aggressive oral corticosteroid therapy given in divided doses is the treatment mainstay. Treatment generally continues over a 2- to 3-year period and is gradually tapered as clinical improvements are seen. The use of steroid-sparing drugs such as methotrexate and oral hydroxychloroquine introduced early in the course of treatment allows for a lower cumulative corticosteroid dose, less potential weight gain, and improvement in growth velocity compared with treatment with corticosteroids only (Feldman et al., 2008).

Patients with JDM often have decreased absorption because of gastrointestinal vasculopathy. Intravenous methylprednisolone has a greater bioavailability than does oral prednisone and may be more effective in patients who have vasculitis in their bowel, causing poor absorption (Wedderburn & Rider, 2009). Infusion of intravenous methylprednisolone may be given once or in repeat doses with low-dose oral prednisone. Adverse reactions may include hypertension, stomach ulceration, anorexia, nausea, ecchymoses, adrenal suppression, hyperglycemia, hypokalemia, thrombophlebitis, weight gain, muscle wasting, Cushingoid appearance, and immunosuppression (i.e., delayed wound healing and increased susceptibility to infection). Use of intravenous methylprednisolone with low-dose oral prednisone may spare some of the untoward adverse effects associated with oral steroids (Feldman et al., 2008).

In addition to its steroid-sparing effects, early introduction of methotrexate (once per week orally or subcutaneously) as an ancillary treatment may improve strength, reduce disease activity, and reduce the incidence of calcinosis (Feldman et al., 2008). Folic acid is administered to prevent folic acid deficiency anemia resulting from adverse effects of methotrexate. Calcium and vitamin D supplements are given prophylactically for bone protection. Hydroxychloroquine, a relatively safe antimalarial drug, may be used to treat the skin rash. It is proposed that hydroxychloroquine has certain mechanisms of action on specific immune cells that activate JDM and that the administration of this drug diminishes symptomatology and is therefore steroid sparing. Hydroxychloroquine adverse effects may include loss of color vision, decreased field of vision, or damage to the retina (Rider, Pachman, Miller, & Bollar, 2007a). Ophthalmologist visits are strongly recommended every 6 months for patients taking hydroxychloroquine.

Cyclosporin may be added for its steroid-sparing effects as well, but an untoward adverse effect is hirsutism and hypertension (Feldman et al., 2008). Intravenous immunoglobulin has been used for refractory cases, particularly persistent JDM skin rashes (Feldman et al., 2008). Other therapeutic drug options include systemic tacrolimus, azathioprine, mycophenolate mofetil, Rituximab, and cyclophosphamide for severe and refractory disease (Stringer & Feldman, 2006).

Physiotherapy should be initiated early for muscle strengthening and conditioning; it can be safely combined with initial drug treatment (Pilkington & Wedderburn, 2005). Referral to a pediatric rheumatologist who is familiar with JDM should occur as soon as a JDM diagnosis is suspected.

This child's mild muscle weakness was not easily detectable on initial examination. It is not uncommon for the rash to present first with mild muscle weakness that may be missed by the untrained eye. Her mild muscle weakness progressed and ultimately was detected by a pediatric rheumatologist. After a muscle biopsy provided a confirmatory diagnosis, two pulse doses of intravenous methylprednisolone were administered.

TABLE. Diagnostic studies for juvenile dermatomyositis

Diagnostic test	Finding	Significance
Aldolase	11 units/L	Elevated
Albumin/globulin ratio	2.1 units/mL	Elevated
Aspartate aminotransferase	38 units/L	Elevated
Creatine kinase	397 units/L	Elevated
Magnetic resonance imaging of lower extremities	Positive for inflammation	Consistent with juvenile dermatomyositis
Muscle biopsy of left vastus lateralis	Positive for articular inclusions, capillary loss, perivascular chronic inflammatory changes, and perifascicular muscle atrophy	Consistent with juvenile dermatomyositis

Concurrent treatment with oral prednisone and oral hydroxychloroquine was begun. Methotrexate administered subcutaneously once per week and a daily dose of oral folic acid were added approximately 6 weeks later. Methotrexate doses were adjusted throughout the course of treatment. The dosage of oral prednisone was tapered over 6 months. A daily dose of Prevacid and Zofran (as needed) were ordered to manage gastric distress. To protect her bones, she began to take calcium and vitamin D every day. Her muscle inflammation responded well to the initial doses of prednisone, and she regained most of her muscle strength in approximately 4 to 6 weeks. However, her skin rash proved to be refractory, which is not an uncommon occurrence; skin manifestations tend to improve more slowly than muscle strength, and persistent skin inflammation sometimes requires different therapies (Lowry & Pilkington, 2009).

Once she was weaned from prednisone and methotrexate, she experienced a skin flare (with no muscle involvement) that consisted of pruritus, worsening of Gottron's papules, and periungual telangiectasias. Oral mycophenolate mofetil was added to her regimen over a 15-month period. After 4 months of taking mycophenolate mofetil she continued to have skin inflammation, and thus intravenous administration of immunoglobulin was begun. She received 10 g every 4 weeks for 6 months, then every other month for a duration of 12 months. At the end of 12 months, she had no signs of active disease. Tapering of her remaining medications continued without consequence, and she was declared in remission 36 months after initiation of treatment.

CONCLUSION

Children who have had ongoing undiagnosed illness may not meet all the criteria for a diagnosis of JDM. Their muscle enzymes may normalize over time, making muscle inflammatory markers alone a poor indicator of disease activity (Pachman et al., 2006). For this reason, it is *imperative* for the practitioner to understand that a persistent skin rash indicates continued active disease even in the presence of normal enzyme levels and muscle strength. Active disease indicates the need for continued aggressive treatment until all clinical signs of disease activity are gone (Feldman et al., 2008).

It is crucial for the primary care practitioner and various specialists to provide collaborative care. Having one primary care practitioner be at the center of what may become a large circle of specialists is important. This approach will help ensure continuity of interdisciplinary care.

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Addressing the family's emotional needs is important. An initial diagnosis of JDM is frightening, and the family will have many questions. No one can tell the family exactly what to expect or what the duration of the child's illness may be. General guidelines of 2 to 3 years of treatment may be given, but because each child's symptoms present differently and each child responds differently to medication regimens, no absolute parameters may be given. The family not only will be trying to process the immediacy of the needed treatment, but they also may be grieving the loss of the "perfect child."

The importance of taking medications regularly in order to keep the disease at bay should be stressed. Teaching must also involve skills in caring for an immunosuppressed child, such as frequent hand washing and keeping the child away from others who are ill. Immunosuppressive therapy requires serial laboratory work to follow the patient's progress and to ensure that the patient does not become too immunocompromised. The family will need to be educated in this regard.

Because the JDM rash can be photosensitive, regular use of sunscreen with a sun protective factor of at least 30 is recommended. The family also should be instructed

about the importance of wearing photo-protective clothing such as hats and long-sleeved shirts when appropriate (Rider et al., 2007b).

The family should purchase the child a medic alert bracelet. Should the child become acutely ill, it is imperative that medical personnel have access to the child's medical diagnosis and an accurate list of medications.

FAMILY RESOURCES

Families of children who are newly diagnosed with JDM are often overwhelmed. Cure JM is an organization that helps families learn more about the disease and connect with other families who have children with JDM for support. Families should be strongly encouraged to review the Web site at www.CureJM.com. In addition, a JDM diagnosis qualifies the child to participate in the Make-a-Wish program.

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