Skin deep: concerning rashes with severe dyspnea

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Case Presentation

HPI: A 27 y.o. female without significant past medical history who presented with progressive exertional dyspnea and diffusely tender, pruritic rash after an upper respiratory infection.

PMHx:
- None

Medications:
- None

Social History:
- Smoking or Vaping: Never
- Alcohol: No
- Illicit drug: No

Physical Exam:
- Gen: appropriate mood and affect.
- HEENT: no scleral icterus or conjunctival injection. Mouth with MMM w/o ulcers. No cervical lymphadenopathy.
- Lungs: Non-labored RA. CTABL.
- Cardiovascular: RRR w/o murmurs, rubs, or gallops
- Abdomen: SNTND.
- Skin: Frontal hair thinning, erythema over malar cheeks and subtle on eyelid, Gottron sign over b/l elbow, MCPs, PIPs, DIPs, palmar papules on palmar fingers. No ulcers or pitting edema. Mild Holster sign b/l.
- MSK: No arthralgia or synovitis. Sensory to pinprick, light touch, temperature, position, vibration normal. Deep tendon reflexes of upper and lower extremities were normal. No Babinski's or Hoffman sign.
- Muscle strength
  - neck flexion 4/5, extension 5/5, SCM 5/5, Trap 5/5
- Upper extremities
  - Shoulder external rotation 4/5, deltoid 4/5-; otherwise 5/5
- Lower extremities
  - Hip flexion 4/5 (b/l), hip extension 5/5 (b/l), hip abduction 4/5 (bl), adduction 4/5 (b/l); all distal extremity were 5/5

Labs:
- ESR 3.1, CRP 1.1
- CK 42-133, Aldolase 4-5.4
- HIV neg, Hepatitis panel neg
- AST 40-43, ALT 52-62, ALP 158, Tbili 4.12-5.7
- Neg HIV, parvo Ab IgG 6.9
- Rheumatology
  - +ANA (1:640, speckled)
  - Neg ENA, dsDNA, CCP, ANCA
  - C3 and C4 wnl
  - A1AT neg, ASMA neg
  - AURP myositis panel
    - Highly positive MDA5 abx
    - Confirmed by OMRF

Diagnostic
- Skin biopsy – Left medial thigh
  - Minimal patchy perivascular lymphocytic dermatitis
- CT Chest
  - Peripheral and basilar ground glass and reticulation c/f early ILD
- PFT
  - FVC 73%, TLC 76%, DLCO 67%, MEP 44%
  - Mild restrict pattern, possible ILD
- CT Abdomen
  - w/o malignancy

Clinical Course

Nov – Dec 2020
- URI sx
- Erythematous and pruritic rash on knuckles, scalp, chest, abdomen, and thigh
- Dx: atopic dermatitis
- Severe fatigue, no longer could jog

Dermatology
- Skin bx: subtle vacuolar interface dermatitis

January 2021
- Jaundice
- Elevated transaminases, ALP, Tbili
- CK and aldolase wnl

Hepatology
- Abd MRI = normal
- Liver biopsy
- Autoimmune hepatitis

Jan-March 2021
- Raynaud
- Arthralgia
- Severe eSOB -> sedentary

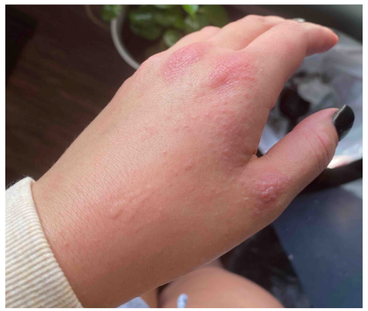
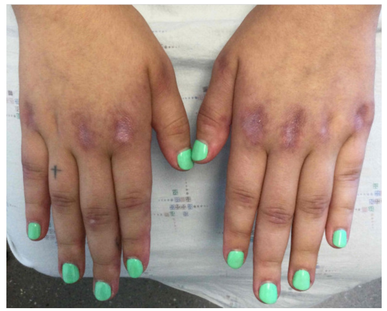
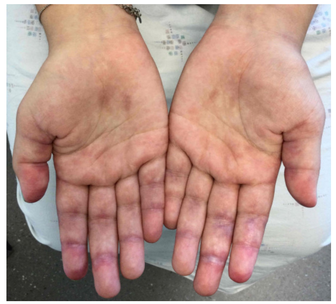
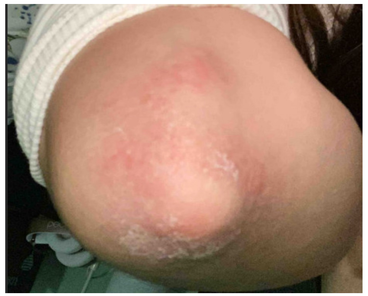
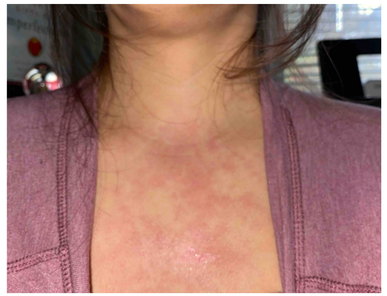
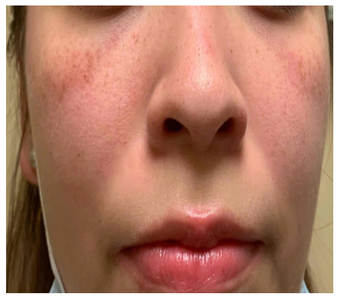
Rheumatology
- ARUP myositis panel positive
  - +MDA5 antibody

Treatment
- Prednisone
- AZA

July 2021
- Rheumatology
- PFT & HRCT
  - Mild restrictive possible ILD
- EMG
  - Diffuse myopathy
- CT abdomen
  - w/o malignancy

Treatment
- MMF
- IVIG

Imaging

Discussion

- Dermatomyositis is an inflammatory myopathy characterized by classic dermatologic findings and symmetrical proximal muscle weakness.

- Cutaneous findings:
  - Gottron papules and Gottron sign
  - Heliotrope rash
  - Shawl or V-sign photosensitive rash
  - Calcinosis cutis
  - Facial erythema (mimic malar rash of SLE)

- Labs:
  - Elevated muscle enzymes (CK, aldolase, CK-MB)
  - Myositis-specific autoantibodies (20-40)
  - Myositis-associated autoantibodies
    - Anti-Ro, anti-La, anti-Sm, or anti-ribonucleoprotein

- MDA5-associated dermatomyositis
  - Positive anti-melanoma differentiation-associated gene 5 (MDA5) antibodies
  - Lower incident of myositis (normal CK and aldolase)
  - Erythematous, painful palmar macules and papules
    - Especially over finger creases and palms
  - Oral ulcers, arthritis, prominent nonscarring alopecia

- Interstitial lung disease
  - 2/3 of MDA+ dermatomyositis cases have ILD
  - 20x higher odd of development compared to MDA5 negative DM
  - *Rapidly* progressive, with hypoxemia within 3 months from respiratory symptom onset
  - Mortality rate: 33-76%

Teaching Points

- MDA5+ dermatomyositis presents with characteristic skin eruption of typical DM; however, may have normal muscle enzyme labs and more specific cutaneous presentation.

- Given the rapidly progressive nature ILD of MDA+ dermatomyositis, prompt diagnosis and aggressive treatment is imperative.

References

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