Diagnostic challenge

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What is the most likely diagnosis of a 2-year-old child with difficulty in walking associated with skin changes on the face and hands?

a) Juvenile systemic lupus erythematosus  
b) Psoriasis  
c) Juvenile dermatomyositis  
d) Juvenile polymyositis  
e) Atopic dermatitis

Juvenile dermatomyositis is the most common idiopathic inflammatory myopathy in childhood. It is characterized by skin lesions associated with symmetric proximal muscle weakness. It is more common in females aged 4-10 years. Onset before the age of 2 years is uncommon.

Muscular involvement is mainly observed in the pelvic and shoulder girdles, resulting in falls and difficulty in walking, climbing stairs, getting up from the chair, reaching objects, etc. Typical skin lesions include a peripalpebral erythematous-violaceous edema (heliotrope), which may extend over the bridge of the nose and malar regions, covering the entire face, as well as reddish papules on the extensor surfaces of the metacarpophalangeal and interphalangeal joints, evolving to depigmented and atrophic lesions (Gottron papules).

Similar changes may occur on the extensor surfaces of the knees, elbows, and ankles (the so-called Gottron’s sign). Another typical cutaneous manifestation is photosensitive erythema, which may lead to the V-sign that occurs in the anterior region of the neck and anterior chest; another manifestation is the shawl sign, which is observed in the neck, shoulders, and side of the arms.

The differential diagnoses of these skin lesions include systemic lupus erythematosus, facial erythema, and psoriasis, according to the location of the Gottron’s papules. Bohan and Peter’s diagnostic criteria for juvenile dermatomyositis consider not only skin lesions and muscle weakness but also the increase in muscle enzymes, evidence of myositis by electromyography, and muscle biopsy changes. In practice, biopsy is reserved for cases with uncertain diagnosis or insufficient therapeutic response. For this reason, patients are usually diagnosed with probable dermatomyositis, as dermatomyositis can only be defined through biopsy.

Despite its low incidence, dermatomyositis usually presents in characteristic form; early diagnosis by the general pediatrician is essential to avoid treatment delay. Delays in diagnosis and treatment can lead to consequences such as lipodystrophy, loss of function and muscle mass, calcinosis, joint contractures, and extramuscular involvement.

REFERENCES