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"LATE ONSET, EARLY IMPACT: THE SUBTLE PROGRESSION OF POMPE DISEASE"

Neurology

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KEYWORDS

A 24-year-old male, with normal birth and developmental history, presented with insidious onset and gradually progressive weakness in the lower and upper limbs over the past five years. The initial symptoms were difficulty climbing stairs, getting up from a squatting position, and increasing difficulty with walking. The symptoms progressively worsened, requiring one-hand support to rise from a seated position. He also described muscle cramps after long-distance walking, but there was no second wind phenomenon or red-colored urine.

The patient reported difficulty with raising his arms above the shoulder and neck weakness, requiring support to lift his head from the bed. Additionally, he had difficulty turning in bed without assistance. He experienced exertional dyspnea and occasional orthopnea. The patient also had a significant family history, with an elder sister's early death attributed to abdominal pain and sudden death, which raised suspicion for a genetic or metabolic disorder.

Clinical Examination:
On examination, the patient had proximal and distal weakness in both the upper and lower limbs. There was marked weakness in neck flexion, with relatively spared neck extension. Other notable findings included:

- Asymmetric scapular winging and secondary axillary folds
- Medial thigh muscle wasting
- Asymmetric calf atrophy
- Preserved deltoid, biceps, and brachioradialis strength
- Preserved extensor digitorum brevis (EDB) function
- Paradoxical breathing (due to intercostal muscle weakness)
- Abdominal muscle weakness

The patient's neck, truncal, and respiratory muscle weakness suggested a progressive, generalized myopathy, potentially affecting both proximal and distal muscle groups.

Differential Diagnosis:
Given the clinical features, the following differential diagnoses were considered:

- **Late-Onset Pompe Disease (Glycogen Storage Disease Type II):** The patient's presentation of progressive proximal muscle weakness, respiratory insufficiency, and a family history suggestive of a genetic disorder aligns with Pompe disease. Pompe disease, caused by acid alpha-glucosidase deficiency (GAA), can lead to glycogen accumulation in muscles and progressive weakness, including respiratory failure in late-onset forms.
- **Facioscapulohumeral Muscular Dystrophy (FSHD)**: FSHD is characterized by facial, scapular, and lower limb muscle weakness, often with a distinctive asymmetric pattern. Neck and truncal involvement, along with scapular winging, raised suspicion of FSHD. However, the lack of facial weakness and the progression of respiratory symptoms suggested that Pompe disease might be a more likely diagnosis.

Investigations:
NCV and EMG were performed, suggesting myopathy.
MRI of the muscles with contrast was performed
Enzyme analysis for Pompe disease showed reduced lysosomal acid alpha-glucosidase activity.
Muscle biopsy did not show any vacuoles with glycogen deposition or features suggestive of other metabolic disorders or dystrophies.

DISCUSSION:
This case highlights the importance of considering late-onset Pompe disease in the differential diagnosis of progressive muscle weakness, especially in patients with respiratory involvement and a family history of early death suggestive of a genetic or metabolic disorder. The pattern of muscle weakness, respiratory symptoms, and positive family history are all suggestive of Pompe disease, though FSHD remains a differential diagnosis due to the asymmetry of muscle involvement and the presence of neck and truncal weakness.

Early diagnosis and enzyme replacement therapy (ERT) have been shown to significantly improve outcomes in Pompe disease, including muscle strength, respiratory function, and overall quality of life. Genetic testing and enzyme analysis remain the cornerstones of diagnosing Pompe disease, while muscle biopsy and MRI can provide additional insights into muscle pathology and confirm the clinical suspicion.

CONCLUSION:
This case emphasizes the importance of a thorough clinical assessment, detailed family history, and appropriate diagnostic workup in patients presenting with progressive muscle weakness. Pompe disease should be considered as a potential diagnosis in patients with proximal muscle weakness, respiratory insufficiency, and a positive family history. Early diagnosis and treatment with enzyme replacement therapy can significantly improve the patient's quality of life and overall prognosis