Electronic Medical Record (EMR) Summary

Patient ID: PID72205020

Name: Meena

Age: 1, Sex: Female

Visit ID: VISIT77628465

Date: 2025-05-17 15:15

Clinical Reasoning Summary

Definition & Key Concerns

The case likely represents a neuromuscular disorder, possibly a form of muscular dystrophy. The delayed walking, frequent falls, difficulty standing up from the floor (Gower's sign), and hypertrophy of the calf muscles are all suggestive of this.

Differential Diagnosis

1. Duchenne Muscular Dystrophy (DMD): This is the most likely diagnosis given the patient's symptoms. DMD is an X-linked recessive disorder characterized by progressive muscle weakness and hypertrophy of the calf muscles. It typically presents in early childhood with delayed motor milestones, frequent falls, and difficulty rising from the floor (Gower's sign).

2. Becker Muscular Dystrophy: This is a milder form of DMD and could also present with similar symptoms. However, it typically presents later in childhood or early adolescence.

3. Spinal Muscular Atrophy: This is a less likely possibility as it usually presents with weakness and atrophy, not hypertrophy, of the muscles.

Can?t-Miss Diagnosis

Duchenne Muscular Dystrophy is the can't-miss diagnosis in this case due to its progressive nature and potential for severe disability and early mortality if not managed appropriately.

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Suggested Investigations

Initial blood tests should include a complete blood count, electrolytes, liver function tests, and creatine kinase (CK) level. A significantly elevated CK level is suggestive of a muscular dystrophy. Genetic testing for mutations in the dystrophin gene can confirm the diagnosis of DMD or Becker Muscular Dystrophy. A muscle biopsy may also be considered if genetic testing is inconclusive.

Management Plan

Management of DMD is primarily supportive and includes physical therapy to maintain mobility and prevent contractures, respiratory support as needed, and corticosteroids to slow the progression of muscle weakness. Cardiac management is also important due to the risk of cardiomyopathy.

Reference Insight

According to UpToDate 2023, early diagnosis and management of DMD can significantly improve quality of life and survival. Genetic counseling is also recommended for families with a history of DMD or Becker Muscular Dystrophy.

Rare Disease Alerts

None triggered

Prescription

None provided