Electronic Medical Record (EMR) Summary

Patient ID: PID60225863

Name: Aarav Kumar

Age: 3, Sex: Male

Visit ID: VISIT13004205

Date: 2025-05-17 16:33

Clinical Reasoning Summary

Definition & Key Concerns

The patient's clinical presentation of delayed walking, frequent falls, difficulty standing from the floor (Gower's sign), and hypertrophic calf muscles is suggestive of a neuromuscular disorder. Duchenne Muscular Dystrophy (DMD) is a primary concern given the age, gender, and symptoms. DMD is an X-linked recessive disorder characterized by progressive muscle weakness and wasting.

Differential Diagnosis

1. **Duchenne Muscular Dystrophy (DMD)**: The most likely diagnosis given the Gower's sign, calf pseudohypertrophy, and delayed motor milestones. DMD typically presents in boys between 1-3 years of age.
2. **Becker Muscular Dystrophy (BMD)**: Similar to DMD but with a later onset and slower progression.
3. **Spinal Muscular Atrophy (SMA)**: This could present with similar symptoms, but calf pseudohypertrophy is not typically seen.
4. **Congenital Myopathy**: These are rare disorders that can present with muscle weakness, but they usually manifest at birth or in early infancy.
5. **Limb-Girdle Muscular Dystrophy (LGMD)**: A group of disorders that cause weakness and wasting of the proximal muscles of the arms and legs, typically presenting in late childhood or early adulthood.

Can?t-Miss Diagnosis

Duchenne Muscular Dystrophy is the critical high-risk condition that must be ruled out given the patient's symptoms and age.

Suggested Investigations

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1. **Creatine Kinase (CK) Level**: This should be significantly elevated in DMD.
2. **Genetic Testing**: Confirmatory test for DMD, BMD, and SMA.
3. **Electromyography (EMG) and Nerve Conduction Studies (NCS)**: Can help differentiate between myopathy and neuropathy.
4. **Muscle Biopsy**: If genetic testing is inconclusive, a muscle biopsy can be performed.

Management Plan

Management of DMD is primarily supportive and includes physical therapy, occupational therapy, and use of assistive devices as needed. Corticosteroids (prednisone 0.75 mg/kg/day or deflazacort 0.9 mg/kg/day) can be used to slow the progression of muscle weakness. Cardiac and respiratory function should be monitored closely.

Reference Insight

According to UpToDate 2023, the diagnosis of DMD is based on clinical findings, elevated serum CK levels, and genetic testing. Early diagnosis allows for timely initiation of corticosteroid therapy, which can slow the progression of muscle weakness and delay the onset of disability.

Rare Disease Alerts

Duchenne Muscular Dystrophy (matched 3 symptoms)

Prescription

None provided