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 case likely represents a neuromuscular disorder, specifically a form of muscular dystrophy. The delayed walking, frequent falls, difficulty standing up from the floor (Gower's sign), and hypertrophic calf muscles are classic signs of Duchenne Muscular Dystrophy (DMD).

Differential Diagnosis

1. **Duchenne Muscular Dystrophy (DMD)**: The most likely diagnosis given the age of onset, Gower's sign, and calf pseudohypertrophy. DMD is an X-linked recessive disorder that primarily affects boys.
2. **Becker Muscular Dystrophy (BMD)**: Similar to DMD but with a later onset and slower progression. However, the patient's symptoms seem to be severe for BMD.
3. **Spinal Muscular Atrophy (SMA)**: This condition can cause muscle weakness and delayed motor milestones. However, it typically presents with weakness of proximal muscles without hypertrophy.
4. **Congenital Myopathy**: These are a group of disorders that can cause delayed motor milestones and muscle weakness. However, they are usually associated with hypotonia and do not typically cause muscle hypertrophy.
5. **Limb-Girdle Muscular Dystrophy (LGMD)**: This is a less likely diagnosis due to the patient's age and pattern of muscle weakness.

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 complications, including respiratory and cardiac failure.

Suggested Investigations

1. **Creatine Kinase (CK) Level**: This is often significantly elevated in DMD.
2. **Genetic Testing**: Confirmation of DMD requires identification of a mutation in the DMD gene.
3. **Muscle Biopsy**: This can show characteristic findings of DMD, but is usually not necessary if genetic testing is available.

Management Plan

Management of DMD is primarily supportive and includes:

1. **Physical Therapy**: To maintain mobility and prevent contractures.
2. **Steroids**: Prednisone or Deflazacort can slow the progression of muscle weakness.
3. **Cardiac and Respiratory Management**: Regular monitoring and early intervention for cardiac and respiratory complications is crucial.

Reference Insight

According to UpToDate (2023), the diagnosis of DMD is typically suspected based on clinical findings and elevated CK levels, and confirmed by genetic testing. Management is multidisciplinary and aims to maintain function, manage complications, and improve quality of life.

Rare Disease Alerts

Duchenne Muscular Dystrophy (matched 4 symptoms)

Prescription

None provided