

Electronic Medical Record (EMR) Summary

Patient ID: PID60225863

Name: Aarav Kumar

Age: 3, Sex: Male

Visit ID: VISIT64076155

Date: 2025-05-17 14:24

Clinical Reasoning Summary

****Definition & Key Concerns****

The child's presentation of delayed walking, frequent falls, difficulty standing up from the floor (Gower's sign), and hypertrophy of the calf muscles are suggestive of a neuromuscular disorder, likely Duchenne Muscular Dystrophy (DMD). DMD is a genetic disorder characterized by progressive muscle degeneration and weakness due to alterations in the dystrophin gene.

****Differential Diagnosis****

1. Duchenne Muscular Dystrophy (DMD): The most likely diagnosis given the child's age, sex, and the presence of Gower's sign and calf hypertrophy. DMD typically presents around age 2-3 with delayed motor milestones and progressive muscle weakness.
2. Becker Muscular Dystrophy: Similar to DMD but typically presents later and progresses more slowly. However, early-onset cases can occur.
3. Spinal Muscular Atrophy: This condition can cause muscle weakness and delayed motor milestones, but it is usually associated with other symptoms such as respiratory difficulties.
4. Congenital Myopathies: These are a group of muscle disorders that can cause hypotonia and muscle weakness, but they usually present at birth or in infancy.

****Can't-Miss Diagnosis****

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Duchenne Muscular Dystrophy is the critical high-risk condition that must be ruled out given the patient's symptoms and the progressive, debilitating nature of the disease.

****Suggested Investigations****

1. Serum Creatine Kinase (CK) Level: This is often markedly elevated in DMD due to muscle breakdown.
2. Genetic Testing: Confirmation of DMD is made by identifying mutations in the dystrophin gene.
3. Muscle Biopsy: If genetic testing is inconclusive, a muscle biopsy showing absence or reduction of dystrophin can confirm the diagnosis.

****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 treatment of cardiac and respiratory complications is necessary as the disease progresses.

****Reference Insight****

According to UpToDate 2023, early diagnosis of DMD is crucial as it allows for early initiation of treatment and genetic counseling for the family. Genetic testing is the preferred diagnostic method, but CK levels and muscle biopsy can also be used. Management is primarily supportive and aims to maintain function and quality of life.

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Rare Disease Alerts

None triggered

Prescription

None provided