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Physical Therapy Intervention for Spinal Muscular Atrophy

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Abstract: ***This case report describes a 12-year-old male presenting with a 1-year history of progressive inability to walk and stand without support, following a period of complete recovery from tuberculosis meningitis at age 3. Functional assessment reveals significant limitations in mobility, including inability to roll independently, walk, or sit without support. Physical examination demonstrates generalized muscle weakness (predominantly 1/5 and 2/5 on MMT in lower and upper limbs respectively), normal tone, and intact sensations. Deep tendon reflexes are diminished to absent in the lower limbs. The diagnosis of Spinal Muscular Atrophy (SMA) is suspected based on the clinical presentation. A comprehensive physical therapy treatment plan focusing on improving activities of daily living, chest mobility, preventing complications, and improving strength and endurance is outlined. This case highlights the challenges in managing progressive neuromuscular disorders and the critical role of rehabilitation in optimizing function and quality of life.***

Keywords: Spino Muscular Atrophy, Assistive device

1. Introduction

Patient Information
A 12-year-old male, Poonam Chandra (pseudonym for anonymity), a school-going boy in 2nd standard, presented with the chief complaint of inability to walk since 1 year prior to presentation. He also reported being unable to stand without support. The informant was his father.

History of Present Illness
The patient was reportedly asymptomatic until 3 years prior to presentation, when he was diagnosed with tuberculosis (TB) meningitis. He received hospitalization and medication for 6 months, after which he was stated to have made a complete recovery. However, approximately 1 year prior to the current presentation, his parents noticed a gradual onset of weakness in his lower limbs, manifesting as frequent falls during walking. This weakness progressed, culminating in the inability to walk or stand without support. The family sought medical attention on August 22, 2012.

Past Medical History: Significant past medical history includes TB meningitis at 3 years of age, with reported complete recovery. There is no other significant medical history or hospitalizations of relevance.

Developmental History: The patient reportedly achieved all developmental milestones at normal time and age.

Immunization History: Immunization history is not remembered by the parents.

Socioeconomic Status: Poor.

Pedigree Chart: The provided pedigree chart suggests a familial pattern of disease. The parents are unaffected, and the patient has two younger siblings (11 years and 9 years old) who are also unaffected. However, one older male sibling died at 13 years of age due to TB Meningitis. This familial history is relevant and might suggest a genetic predisposition or a shared environmental factor, though the current patient's progressive weakness points more towards a neuromuscular disorder.

2. Observation and Examination

Upon observation, the patient was well co-operative and oriented to time, place, and person. Higher mental functions appeared normal, with the patient understanding, able to speak, and recognizing parents.

Cranial Nerve Examination:
- Vision: Able to recognize size, shape, and color; focusing on striking objects purposefully. No squint, no facial asymmetry. Light reflex intact. All cranial nerves were intact.

Chest Examination:
- Breathing Pattern: Abdomino-thoracic.
- Chest Excursion: No apparent chest excursion, but kyphosis noted.
- Bilateral air entry equal.

Upper Limb Examination:
- **Motor Examination:**
  - **Tone:** Normal in both upper limbs.
  - **Passive ROM:** Full and free, with no contractures or deformities.
  - **Deep Tendon Reflexes (DTRs):** Biceps (Right: NE, Left: NE), Triceps (Right: NE, Left: NE), Brachioradialis (Right: NE, Left: NE). (NE = Not Elicitable)
  - **Manual Muscle Testing (MMT):**
    - Shoulder Flexors: RT 2+, LT 2+
    - Shoulder Extensors: RT 2+, LT 2+
    - Shoulder Abductors: RT 2+, LT 2+
    - Shoulder Adductors: RT 2+, LT 2+
    - Elbow Flexors: RT 2+, LT 2+
    - Elbow Extensors: RT 2+, LT 2+
    - Wrist Flexors: RT 2+, LT 2+
    - Wrist Extensors: RT 2+, LT 2+
- **Sensory Examination:** All sensations were intact.

Lower Limb Examination:
- **Tone:** Normal.
- **Deep Tendon Reflexes (DTRs):** Knee (Right: +, Left: +), Ankle (Right: +, Left: +), Plantar (Right: NE, Left: NE). (Note: Discrepancy with Upper limb DTRs, where they were NE, here they are + for Knee and Ankle. This should be clarified for publication.)
- **Passive ROM:** Full and free, no contractures or deformities. (Note: Contradicts finding of "tightness of hamstrings B/L" later in the notes.)
- **Manual Muscle Testing (MMT):**
  - Hip Flexors: RT 2+, LT 2+
  - Hip Extensors: RT 1, LT 1
  - Hip Abductors: RT 2+, LT 2+
  - Hip Adductors: RT 2+, LT 2+
  - Knee Flexors: RT 1+, LT 1+
  - Knee Extensors: RT 1+, LT 1+
  - Ankle Plantarflexors: RT 1, LT 1
  - Ankle Dorsiflexors: RT 1, LT 1
- **Other Findings:** Tightness of hamstrings bilaterally.

Functional Examination:
- Unable to roll independently.
- Not able to walk.
- Unable to stand without support.
- Able to sit with support.

Diagnosis: Spinal Muscular Atrophy (SMA) suspected.

Physical Therapy Treatment (PT Treatment):

Goals:
- To improve Activities of Daily Living (ADL) activity.
- To improve chest mobility.
- To make him functionally independent.
- To prevent complications (e.g., contractures, respiratory issues).
- To improve strength.
- To improve endurance.
- To teach energy-saving techniques.

Physical Therapy Rehabilitation Orders (PT Rx):
- **Upright Mobility:** KAFO (Knee-Ankle-Foot Orthosis) is advised if needed. Crutches will be given once independent upright mobility is achieved.
- **Seated Mobility:** This indicates a focus on maximizing independent movement while seated.
- **Strengthening Exercises:** Specific exercises targeting weak muscle groups, likely focusing on residual strength in a gravity-eliminated or assistive environment.
- **Positioning:** To prevent contractures and pressure sores.
- **Energy Saving Techniques:** Strategies to conserve energy during daily activities, given muscle weakness.
- **Chest Expansion Exercises:** To address chest kyphosis and improve respiratory function.

3. Discussion

Spinal Muscular Atrophy is a genetic neuromuscular disorder characterized by degeneration of alpha motor neurons in the spinal cord.[1] This case highlights the progressive nature of suspected Spinal Muscular Atrophy in a 12-year-old male. The history of normal early development followed by insidious onset of weakness, especially after a significant medical event (TB meningitis), is crucial. While TB meningitis itself can lead to neurological sequelae, the pattern of progressive, symmetrical weakness, particularly in a familial context (older sibling died of TB meningitis, suggesting a potential shared genetic or environmental vulnerability, although the direct link to SMA is not confirmed), strongly points towards a primary neuromuscular disorder like SMA. The presence of generalized weakness (more pronounced in lower limbs), diminished/absent DTRs, and intact sensation are classic indicators of a lower motor neuron lesion, consistent with SMA. The noted kyphosis further supports chronic muscle weakness affecting spinal posture. Physical therapy plays a crucial role in maintaining functional ability and preventing contractures in children with SMA[2]

The functional limitations are significant, impacting basic mobility like rolling, sitting, standing, and walking. The physical therapy goals are appropriately focused on improving ADLs, preventing secondary complications such as contractures and respiratory compromise, and maximizing residual strength and endurance.

Early rehabilitation intervention is vital to delay functional deterioration and improve quality of life[3] Chest physiotherapy helps prevent respiratory complications in patients with neuromuscular diseases[4]

The recommendation of KAFOs and crutches indicates a strategy to provide external support for upright mobility, which is critical for participation and quality of life in patients with progressive weakness. Orthotic support such as KAFOs can enhance mobility and upright tolerance.[5] Energy-saving techniques are paramount to prevent fatigue and enable participation in daily activities.

4. Limitations

A definitive diagnosis of SMA requires genetic testing. The current notes only provide a "suspected" diagnosis. Further investigations, including electromyography (EMG), nerve conduction studies (NCS), and genetic testing for *SMN1* gene deletion, would be essential to confirm the diagnosis and classify the type of SMA. Long-term follow-up and detailed functional outcome measures would also be beneficial for publication. The discrepancies in DTRs and ROM findings should be clarified.

5. Conclusion

This case report illustrates the presentation and preliminary physical therapy management of a 12-year-old male with suspected Spinal Muscular Atrophy, characterized by progressive muscle weakness and significant functional limitations. Comprehensive rehabilitation, including strengthening, mobility training, and energy conservation, is vital in optimizing functional independence and preventing complications in these patients. This case underscores the importance of a detailed clinical evaluation and the need for genetic confirmation for definitive diagnosis and appropriate long-term management in suspected neuromuscular disorders.

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