E-Poster

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Rare but Real: Rehabilitation Challenges in Vanishing White Matter Disease

Topic: Specific Target Groups (Children, Women, Elderly, Athletes, Musicians, Refugees, Ethnic Groups, Workers, and Others)

Ana Almeida Vieira, Mónica Jorge, Cristina Marques Baptista, João Capelo, Virginia Reis, Isabel Batalha

Department of Physical Medicine and Rehabilitation, Centro de Medicina de Reabilitação de Alcoitão, Lisbon, Portugal

BACKGROUND

Vanishing White Matter Disease (VWMD) is a rare autosomal recessive leukodystrophy caused by mutations in the EIF2B genes, leading to progressive neurological deterioration. It primarily affects the white matter, with symptoms worsened by physiological and emotional stressors. This case highlights the rehabilitation challenges in managing a surviving sibling, particularly regarding motor, respiratory, and swallowing function.

CASE DESCRIPTION

The patients belong to a non-consanguineous family from a developing African country, with no relevant family history. Among three siblings, the eldest and youngest were diagnosed with VWMD, while the middle sibling remains neurologically healthy. Limited healthcare resources delayed diagnosis and access to specialized rehabilitation.

The first affected sibling developed developmental regression at 2 years, presenting with ataxia, spasticity, and cognitive decline. Disease progression was rapid, leading to severe neurological impairment and death at 4 years old. The second sibling exhibited similar early symptoms but with a slower progression, allowing for a prolonged rehabilitation approach. However, her condition remains unpredictable, requiring continuous adaptation of strategies.

Rehabilitation efforts focus on mobility preservation, spasticity management, and functional independence. As the disease advances, respiratory and swallowing dysfunction emerge as critical concerns, with high risks of aspiration pneumonia and respiratory failure. This necessitates early intervention in dysphagia management and respiratory physiotherapy to balance function preservation with medical stability. Conventional rehabilitation must be adjusted to prevent overexertion and deterioration.

Diagnosis was confirmed through brain MRI (diffuse leukodystrophy) and EIF2B genetic testing.

DISCUSSION

Vanishing White Matter Disease presents significant rehabilitation barriers, requiring a dynamic reassessment of therapy goals. Beyond motor decline, swallowing and respiratory complications demand early, multidisciplinary intervention. Anticipatory guidance, adaptive strategies, and careful therapy calibration are crucial to maximizing quality of life while preventing complications.

CONCLUSION

This case underscores the complexity of rehabilitation in VWMD, where balancing motor function, respiratory care, and dysphagia management is essential. A highly individualized, multidisciplinary approach is necessary to optimize patient outcomes while minimizing deterioration risks.

Statement of Disclosures

None

Corresponding Author:
Ana Almeida Vieira
Department of Physical Medicine and Rehabilitation, Centro de Medicina de Reabilitação de Alcoitão, Lisbon, Portugal
Email: anaritavieira97@hotmail.com

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