E-Poster

EP-350

Isaac's Syndrome: A Multimodal Rehabilitation Approach for a Rare Neuromuscular Disorder

Topic: Health Conditions (Nervous System, Musculoskeletal System, Cardiopulmonary System)

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BACKGROUND

Isaac's syndrome (IS), also known as acquired neuromyotonia, is a rare neurological disorder characterized by persistent peripheral nerve hyperexcitability. With an estimated prevalence of 1–2 cases per million, it primarily affects middle-aged males. Key symptoms include continuous muscle twitching, stiffness, cramping, fasciculations, myokymia, pseudomyotonia, rapid heart rate and hyperhidrosis. Rehabilitation medicine plays a crucial role in symptom management, improving motor function, alleviating stiffness, and enhancing overall quality of life. This case report highlights the benefits of a comprehensive rehabilitation program in an adolescent patient with Isaac's syndrome.

CASE DESCRIPTION

We present the case of a 17-year-old male diagnosed with Isaac's syndrome two years prior, who sought medical attention due to bilateral lower limb tremors, more pronounced on the left side, cephalic tremor and intermittent left upper limb tremor. Additional symptoms included gait disorders, fine motor difficulties, and sleep disorders. The patient underwent an extensive diagnostic workup, including genetic, neurological, and cardiological evaluations, as well as radiological imaging, laboratory testing, nerve conduction studies, and electromyography (EMG). A comprehensive assessment of his medical history following a thorough clinical examination guided the development of an individualized rehabilitation program, integrating robot-assisted therapy, virtual reality therapy, occupational therapy, kinesiotherapy, and physiotherapy. The rehabilitation protocol consisted of three hours of daily therapy over a two-week period.

DISCUSSION

Functional status, disability level, quality of life, and psychological impact were systematically assessed at admission and discharge using validated tools, including the Modified Ashworth Scale (MAS), Medical Research Council (MRC) Scale, Modified Rankin Scale (mRS), Pediatric Quality of Life Inventory (PedsQL), Screen for Child Anxiety Related Emotional Disorders (SCARED), Timed Up and Go (TUG) Test, Fatigue Severity Scale (FSS), and Visual Analog Scale (VAS).

Following completion of the rehabilitation program, significant improvements were observed in muscle strength, gait stability, and exercise tolerance. The frequency and duration of myoclonic episodes decreased, while sleep quality and overall well-being improved substantially.

CONCLUSION

A multidisciplinary rehabilitation approach is a crucial component in managing Isaac's syndrome in adolescent patients. The goals of the rehabilitation program are to reduce neuromuscular hyperexcitability, prevent rigidity, and optimize functional status, thereby facilitating long-term improvements in mobility, enhancing quality of life, and ensuring long-term functional independence.

Statement of Disclosures

None

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Doi: 10.5606/archisprm.2026.EP-350