ABSTRACT

Background

Congenital myopathies represent a heterogeneous group of genetic skeletal muscle diseases characterised by specific histopathological alterations. Among these, Central Core Myopathy (CCM) associated with mutations in the RYR1 gene is one of the most common forms, often with early onset and varying severity. The main symptoms include hypotonia, arthrogryposis, respiratory failure, and skeletal deformities.

Despite the importance of multidisciplinary care, scientific literature offers little evidence on the effectiveness of physiotherapy and therapeutic exercise in children with this condition.

Objectives

The aim of the study is to evaluate, through two clinical cases of heterozygous twins in paediatric age diagnosed with Central Core congenital myopathy and arthrogryposis, the effectiveness of continuous physiotherapy treatment in improving neuromotor skills and to compare the functional evolution of the two subjects.

Methods

The study consists of two case reports written in accordance with CARE guidelines and conducted at a local Rehabilitation Medicine and Physiatry Unit between November 2024 and September 2025, after obtaining the necessary authorizations from the health authority involved.

Two heterozygous twin patients with homozygous RYR1 Central Core Myopathy with arthrogryposis were included. Both underwent individualised, continuous physiotherapy treatment twice a week for nine months.

Assessments were performed in three stages (November 2024, April 2025, July 2025) using the *Hammersmith Functional Motor Scale Expanded (HFMSE)*, the *CHOP Intend* and the joint PROM assessment in the areas most affected by arthrogryposis.

Results

Both patients showed progressive improvement in motor skills and participation in activities, with differences related to clinical severity.

In case 1 (N.S.), with tracheostomy and J-PEG, there were increases in head control and proximal motor skills and greater participation in the proposed activities.

In case 2 (E.S.), in better clinical conditions, progress was more marked, with improved postural control, increased *HFMSE* and *CHOP Intend* scores, and greater interaction and communication.

In both cases, physiotherapy contributed to maintaining joint mobility and preventing contractures.

Conclusion

Early and continuous physiotherapy treatment has shown positive effects on motor function and, consequently, on quality of life, highlighting the central role of physiotherapy in the management of children with congenital myopathy.

The different development of the twins highlights the importance of an individualised approach that is attentive to the characteristics of each child.

Although the results are limited by the small sample size, the study supports the need for further research to define standardised and validated rehabilitation protocols for this category of patients.

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