

ABSTRACT - Caterina Sirocchi

Background

PHACE syndrome is a rare neurocutaneous disorder characterized by the association of posterior cranial fossa abnormalities (P), infantile facial hemangiomas (H), arterial abnormalities, especially aortic (A), cardiac abnormalities (C) and ocular abnormalities (E). It presents itself with extremely heterogeneous manifestations which over time can also lead to serious complications, so it requires multidisciplinary management and continuous follow-up over time.

The study was conducted on a pediatric patient with PHACE syndrome with an ataxic disorder of walking, posture and gesture. It was decided to focus on gesture ataxia, proposing a rehabilitation treatment protocol for the upper limb and evaluating its effectiveness.

Objectives

The aim of the study is twofold: on one side, to question the available literature to understand the different manifestations of PHACE syndrome; on the other, to evaluate the effectiveness of physiotherapy treatment focused on manual skills in a pediatric patient with ataxia in PHACE syndrome.

Methods

A review of the scientific literature on the main databases (Pubmed, Cinhal, Medline) was conducted. This study is a *Case Report* type, compliant with the CARE guidelines. A quarterly treatment protocol, lasting a total of 25 hours, was performed in a pediatric patient with gesture ataxia in PHACE syndrome. The choice of manual skills to be trained (precision, dexterity and calibration) and treatment efficacy were agreed with the family and demonstrated via structured assessment scales administered pre-treatment (T0), post-treatment (T1, after 3 months) and post-washout (T3, after 8 months). The rating scales used are the *Melbourne Assessment 2*, the *9-Hole Peg Test*, structured time trials and the *Goal Attainment Scaling*.

Results

The assessment scales showed significant improvements after the intensive treatment period, with an overall bilateral increase of 12% at Melbourne Assessment 2, while after the washout period there was predominantly a maintenance of acquired skills. Functional improvements in daily living skills are also reported, assessed using the Goal Attainment Scaling.

Conclusion

This study allowed us to investigate the information available in the literature on PHACE syndrome and possible physiotherapy treatments of gesture in pediatric patients with cerebellar ataxia. These results were compared with those that emerged from the clinical case in question, highlighting the lack of studies present in the literature and the different rehabilitation approaches used. In the conclusion, the limitations of this case study and the development proposals for future studies were indicated.

References

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