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Charcot-Marie-Tooth foot in children: Early diagnosis and orthopedic management

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ABSTRACT

Charcot-Marie-Tooth disease (CMT) is an inherited neuropathy that affects the peripheral nerves, resulting in foot deformities and impaired mobility in children. Early diagnosis is essential to prevent the disease from worsening and allow orthopedic interventions that improve function and quality of life. This study reviews the initial clinical manifestations of CMT, diagnostic approaches, and the main orthopedic management strategies, highlighting the importance of early intervention with the use of orthoses and, when necessary, surgical techniques. The review also explores the prospects for functional improvement and symptomatic relief with appropriate treatment.

Keywords: Charcot-Marie-Tooth, Foot deformities, Children, Orthopedic management, Early diagnosis.

INTRODUCTION

Charcot-Marie-Tooth disease (CMT) is one of the most common inherited neuropathies, characterized by progressive degeneration of the peripheral nerves, resulting in muscle weakness, sensory loss, and deformities in the limbs, especially the feet. In children, associated deformities, such as pes cavus and ankle instability, can negatively impact motor development and quality of life, making early diagnosis essential to mitigate disease progression. Orthopedic management, which includes everything from the use of orthoses to surgical interventions, aims to correct deformities and preserve motor function, offering better prognoses for patients. This review addresses the clinical manifestations and main orthopedic treatment strategies for the management of Charcot-Marie-Tooth foot in children, highlighting the importance of early intervention to minimize future complications.

METHODOLOGY

The literature review was systematically conducted in the PubMed, Scopus, and Google Scholar databases, covering studies published between 2015 and 2023. The keywords "Charcot-Marie-Tooth", "foot deformities", "children" and "orthopedic management" were used to search for relevant articles. Inclusion criteria involved clinical studies, systematic reviews, and international guidelines focused on the early diagnosis and orthopedic management of deformities associated with CMT in children. Articles that discussed adults only, non-validated

experimental treatments, or that did not include data on orthopedic interventions were excluded. Data analysis focused on the identification of diagnostic practices, use of orthoses, surgical interventions, and clinical results documented in the selected studies.

This methodological refinement seeks to ensure the review of robust and up-to-date evidence to offer a comprehensive and critical view on current practices in the early diagnosis and orthopedic management of Charcot-Marie-Tooth foot in children.

RESULTS

The reviewed literature demonstrates that the early diagnosis of Charcot-Marie-Tooth disease (CMT) in children depends on a detailed clinical evaluation, identifying signs such as pes cavus, distal muscle weakness, and sensory changes in the lower limbs. Studies suggest that the use of complementary tests, such as electroneuromyography and genetic testing, are effective tools to confirm the diagnosis and classify the subtype of CMT.

With regard to orthopedic management, the application of orthoses is widely recommended in the early stages to promote stability and functionality. In more advanced cases, surgical correction of deformities, such as osteotomy and tendon transfer, has been shown to be effective in maintaining mobility and preventing additional complications, such as ulcers and falls.

The review also highlights the importance of continuous and multidisciplinary follow-up, including physiotherapists and orthopedists, to monitor the progression of the disease and adjust interventions according to the child's clinical evolution.

CONCLUSION

Early diagnosis of Charcot-Marie-Tooth foot in children and proper orthopedic management are crucial to improve quality of life and prevent permanent deformities. The review suggests that the combination of clinical diagnosis and surgical or non-surgical interventions are effective in maintaining foot functionality.

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