CLINICAL AND PARACLINICAL CHARACTERISTICS OF HEREDITARY SPASTIC PARAPLEGIA (STRÜMPELL DISEASE) IN CHILDREN AND OPTIMIZATION OF EARLY DIAGNOSTIC APPROACHES

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ABSTRACT

Hereditary spastic paraplegia (HSP) represents a genetically heterogeneous group of neurodegenerative disorders characterized by progressive lower-limb spasticity, hyperreflexia, and dysfunction of corticospinal tracts. In pediatric practice, HSP is frequently misdiagnosed as spastic diplegic cerebral palsy (CP), largely due to overlapping clinical manifestations at early stages, leading to delayed diagnosis and ineffective or incorrect therapeutic approaches.

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The objective of this study was to investigate the clinical and paraclinical features of Strümpell hereditary spastic paraplegia in children, to evaluate genotypic variants of the SPG gene spectrum, and to optimize early diagnostic algorithms by comparing HSP with spastic diplegic CP.

A total of 153 children were included in the study, comprising 95 patients with HSP, 58 patients with spastic diplegic CP, and 40 healthy controls. A comprehensive diagnostic approach was applied, involving clinical neurological evaluation, assessment of motor function using MRC, Ashworth, and GMFCS scales, MRI of the brain and spinal cord, ENMG of lower-limb nerves, and molecular-genetic investigations including MLPA and next-generation sequencing (NGS) of the SPG4 gene.

The study identified several significant findings. For the first time, a strong correlation was demonstrated between spasticity severity, muscle strength, and patient age in the HSP group, whereas in children with CP the correlation was inverse. Novel pathogenic intronic mutations in the SPG4 gene (chr2:32369901CAT>C and c.1617-105T>C) were detected. Furthermore, an association between complicated forms of HSP and mutations in the

SUPT16H gene was revealed, particularly in patients with structural anomalies of the corpus callosum. The application of NGS enabled early detection of autosomal recessive deletions/duplications in consanguineous families, significantly improving diagnostic accuracy.

Clinical comparison of HSP and CP groups revealed distinct diagnostic markers. HSP was characterized by progressive spasticity, predominant involvement of lower limbs, impaired corticospinal tract conduction, specific MRI signs including spinal cord thinning, and characteristic ENMG changes. A high prevalence of consanguinity and familial cases was observed among HSP patients, underscoring the importance of genetic counseling in affected families.

The study resulted in the development and implementation of an optimized diagnostic algorithm that integrates clinical, neurophysiological, imaging, and genetic criteria. This algorithm has been introduced into clinical practice in several healthcare institutions and has demonstrated effectiveness in improving early diagnosis and management strategies for children with hereditary spastic paraplegia.

Conclusion: A comprehensive diagnostic approach combining clinical assessment with modern genetic testing significantly enhances the early detection and accurate differentiation of hereditary spastic paraplegia from cerebral palsy. The findings contribute to improved clinical management, timely rehabilitation, and effective genetic counseling for families.

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