

عنوان مقاله:

An update on clinical, pathological, diagnostic, and therapeutic perspectives of childhood leukodystrophies

محل انتشار:

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خلاصه مقاله:

Leukodystrophies constitute heterogeneous group of rare heritable disorders primarily affecting the white matter of central nervous system. These conditions are often underappreciated among physicians. The first clinical manifestations of leukodystrophies are often nonspecific and can occur in different ages from neonatal to late adulthood periods. The diagnosis is, therefore, challenging in most cases. Comprehensive evaluation of clinical findings, brain magnetic resonance imaging, and genetic studies play the key roles in the early diagnosis of individuals with leukodystrophies. No cure is available for most heritable white matter disorders but symptomatic treatments can significantly decrease the burden of events. New genetic methods and stem cell transplantation are also under investigation to further increase the quality and duration of life in affected population. The improvements in molecular diagnostic tools allow us to identify the meticulous underlying etiology of leukodystrophies and result in higher diagnostic rates, new classifications of leukodystrophies based on genetic information, and replacement of symptomatic managements with more specific targeted therapies.

کلمات کلیدی:

.Pathological, Diagnostic

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