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LECTURE

Inherited Metabolic Disorders and Stroke

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Abstract

Recent data show that stroke is one of the main causes of death and the main cause of neurological disability in the adult. It is also a major cause of dementia and age related cognitive decline. Conventional cardiovascular risk factors explain a significant percentage of stroke risk, but an important part of that risk remains without explanation. Additionally, different individuals have different outcomes when exposed to the same risk factor. This means that genetic factors have an important role in stroke risk determination. Unlike other cardiovascular diseases, stroke is a heterogeneous disorder and genetic factors can affect stroke pathophysiology at different levels. Taking this into consideration, we can identify several inherited metabolic disorders, usually with multisystemic presentations, that can have stroke as one of their phenotypic expressions. We have disorders that express themselves as small- or large-vessel disease patterns, as cardioembolic causes or other kind of profiles (for instance, stroke-like in MELAS). This group of disorders, although individually rare, are collectively frequent and they can appear at any age, from the newborn to the elderly. Due to their rarity, they are frequently not recognized or taken into account in diagnostic flowcharts, although for a significant number of them we have specific treatments that can improve prognosis significantly.

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