GENETICALLY INHERITED METABOLIC DISORDERS MIMICKING CEREBRAL PALSY

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Cerebral palsy (CP) is an umbrella term encompassing a group of non-progressive multietiological conditions that cause physical disability in human development. The motor impairment can change with age depending on the time of consecutive central nervous system structures and functions maturation in the aftermath of disturbed early stages of brain development. CP is divided into four neurologic syndromes describing the different movement impairments (spastic, dystonic/athetoid, ataxic, mixed). Defects observed in CP, apart from motor dysfunction, are as follows: mental retardation (60%), behavioral disorders (50%), vision impairment (50%), and hearing loss (25%), and well as epilepsy (20-30%).
Genetically inherited metabolic disorders considered as separated entities are not frequently diagnosed but generally they are present in 1 per 500 neonates, and are even seen in every fifth ill term neonate without infectious factor which is not excluded criterion as preterm delivery. Clinical picture is non specific and common for many frequent disorders like preterm delivery and connected with it adaptive problems, infections or hypoxic-ischemic encephalopathy of perinatal period. Similar symptoms seen in consecutive child make the diagnosis of EIM more probable. Owing to introduction of diagnostics strategies-biochemical tests (selective screening), neuroradiological methods and investigations on subcellular-molecular level- rapid progress in that field has been observed.
The authors discuss patients suffered from genetically inherited metabolic disorders (aminoacydopathies, organic acidurias, mitochondrial cytopathies, peroxisomal disorders, neurodegenerative disorders and others), who were previously wrongly diagnosed with spastic or extrapyramidal form of cerebral palsy.