Neuroimaging Findings in Storages Disease of Children

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The diagnosis of metabolic diseases is challenging. Most of them have poor prognosis, leading to early death, whereas milder forms, juvenile and adult live longer. MRI can be of help in some conditions, but in most cases extensive investigations, including genetic tests are needed to reach diagnosis. Several classifications have been proposed for these disorders. They can be classified according to the cell organelle involved, whether it is the lysosome, peroxisome, or mitochondrion, or the underlying biochemical defect. These classifications are, however less helpful because there are no characteristic imaging findings shared by all diseases. The classification to gray and white matter diseases is, although simple and practical and therefore widely used. In some conditions, the involvement of specific white matter tracts can be a diagnostic clue. The deep gray nuclei are affected in many metabolic diseases, also in those with primarily white matter imaging findings, but the results have so far been discouraging because of the large variability of presentations of these disorders. To achieve images of any diagnostic value the patient must be imaged early in the course of the disease.

Niemann-Pick type C disease (NPC) is a rare autosomal recessive lipid storage disorder caused by impaired cellular functions in processing and transport of LDL-cholesterol. The symptoms are usually clumsiness and gait disturbance at the beginning, and include ataxia, mental disturbance, dystonia, dysarthria, dysphagia, seizures and vertical supranuclear gaze palsy during the disease course. Mutations of the NPC1 gene, located on chromosome 18q11-12, are observed in 95% of NPC patients, whereas mutations of the NPC2 gene are noted in 5%. It has been proposed that the mutations in the NPC genes cause blockage of cholesterol transport between lysosomes and the sterol regulator machinery, so that excessive cholesterol is accumulated and apoptosis of neurons occurs. The intracellular accumulation of unesterified cholesterol in the lysosomal system can be detected from the fluorescence level around the nucleus, via filipin staining. To reach a conclusive diagnosis of NPC, both the abnormal accumulation of cholesterol and LDL-cholesterol esterification in fibroblast cell culture should be observed. For NPC, brain MRI usually is not capable of detecting abnormality except in the late stages. MRS the neuroimaging technique for obtaining information about brain metabolism shows decreased N-acetyl aspartate indicates neuronal and axonal loss, whereas increased choline reflects demyelination or gliosis. In addition, 18-fluoro-2-deoxyglucose (F-18 FDG) positron emission tomography (PET), based on glucose utilization in the brain, is known to be sensitive for the detection of metabolic derangements in patients with neurodegenerative disorders.

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