Nieman-Pick Disease Type C: An Epileptologic View

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Niemann-Pick disease type C (NPC) is a lipid storage disease that can present in infants, children, or adults. Neonates can present with ascites and severe liver disease from infiltration of the liver and/or respiratory failure from infiltration of the lungs. Other infants, without liver or pulmonary disease, have hypotonia and developmental delay. The classic presentation occurs in mid-to-late childhood with the insidious onset of ataxia, vertical supranuclear gaze palsy (VSGP), and dementia. Dystonia and seizures are common. Dysarthria and dysphagia eventually become disabling, making oral feeding impossible; death usually occurs in the late second or third decade from aspiration pneumonia. Adults are more likely to present with dementia or psychiatric symptoms.

From epileptologic point of view there a variety of epileptic as well as epileptic like disorders has been described in this disease. From myoclonic seizures to cataplexy has been reported in the literature.

In my short talk, I will address to these manifestations of NPC.

Keywords: NPC; EEG; Pediatric; Epileptology; Seizure; Nonepileptic

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