Juvenile type of Niemann-Pick type C disease and our study in Iranian NPC patients

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The primary biological defect in Niemann-Pick type C disease (NPC) is impaired intracellular lipid transportation that leads to toxic accumulation of lysosomal lipids. In peripheral tissues such as the skin; it is predominantly unesterified cholesterol that accumulates.

In the liver and spleen, several different types of lipid accumulate including unesterified cholesterol, glycosphingolipids, phospholipids and sphingomyelin. Glycosphingolipid accumulation primarily occurs in the central nervous system (CNS).

Vertical supranuclear gaze palsy (VSGP) is the most common neurological sign of NPC and Very strong indicator of this disease.

Ocular-motor abnormalities are one of the earliest neurological signs of NPC and are seen in around 80% of patients. Initial sign is usually impaired voluntary saccadic eye movements (SEM) in the late-infantile period.

Neurological examination of VSGP should include testing of smooth pursuits, saccades and vergence movement of the two eyes in opposite directions. In all patients, saccadic, pursuit and vergence movements should be examined in both vertical and horizontal planes.

Vertical SEM impairment is affected first, followed by horizontal SEM impairment. Gelastic cataplexy is another neurological sign and Very strong indicator of NPC.

It is less common than VSGP, but is a characteristic feature of the disease.

Patients may experience sudden loss of muscle tone (without loss of consciousness), which is typically triggered by an emotional stimulus, especially after laughing. Loss of tone may involve the legs, neck or jaw. Therefore, cataplexy may manifest as sudden falls, sudden head drop or jaw drop.

Ataxia and clumsiness or frequent falls is another neurological manifestation in these patients and it is Moderate indicator of NPC. We may detect poorly coordinated movements affecting walking, which may start at any age from late childhood to adulthood and is progressive.

Dysarthria also is Moderate indicator of NPC Characterized by poor articulation and slurred speech. Dysphasia is Moderate indicator of NPC too. It May be present early in the disease course and May manifest initially as difficulty swallowing liquids. However, during disease progression, increasing dysphagia severity can lead to aspiration and malnutrition

Dystonia also is Moderate indicator of NPC and Can cause abnormal dystonic postures in hands, feet or face.

Epileptic seizures, Most frequent in late-infantile or juvenile-onset NPC patients and May experience different types of seizures, for example, partial/ focal, generalized, absence, myoclonic or tonic-clonic, of variable frequency

and intensity.
Myoclonus may be brief as involuntary twitching of a muscle or a group of muscles. Also these patients may show Progressive hearing loss.
In this article we discuss about neurological sign and symptoms of Juvenile NPC patients.

Keywords: NPC; Juvenile type; Neurological sign