Niemann –Pick C Disease: Is It A Misnomenclature?

How to Cite This Article: Ahmadabadi F. Niemann –Pick C Disease: Is It A Misnomenclature. Iran J Child Neurol. 2015 Autumn;9:4(Suppl.1): 27.

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Niemann-pick Type C is an autosomal recessive disease which is presented by Cholesterol and fatty acid accumulation in liver-Brain and spleen lead to some symptoms.

This Type of disease is etiologically separate from other types of niemann-pick but there are similar symptoms.

The incidence is 1/150000

Neonatal form of disease present with icter, ascites and severe liver disease.

The classic form occurs in childhood with supranuclear gaze palsy (Vertical), Ataxia, Dysarthria and Dystonia.

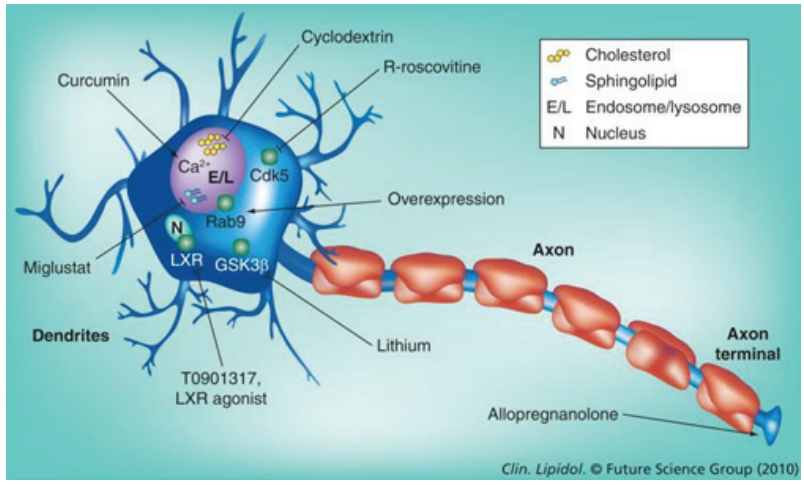
In Adult form Dementia and Psychiatric presentations are common.

Some symptoms as vertical supranuclear gaze palsy and gelastic cataplexy are very diagnostic and others as dysartheria or psychiatric symptoms are seen occasionally.

Diagnosis is based on staining in fibroblasts, Sea blue histiocytes in bone marrow and genetic study.

Substrate inhibition therapy can arrest progression of disease in some patients which is possible with Miglustat (Zaveska) a drug that is used for treatment of Gaucher disease, too.

Keywords: Niemann-pick C; Misnomenclature; Miglustate



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