

Management of NPC

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Nieman-pick type C is a neuro visceral lipid storage disorder. Almost 95% of patients have mutation in NPC1 gene .A smaller group of patients have mutation in NPC2 gene. Both of mutations impair cholesterol-mediated homeostatic responses in the endoplasmic reticulum. Function of NPC1 and NPC2 gene are detected on skin fibroblasts.

Clinical manifestations:

4 phenotypes of NPC are reported:

1- Fetal Type that is presented by splenomegaly and hepatomegaly in utero, utero ascites, intrauterine growth retardation, and oligohydramnios, Placentomegaly and intervillous thrombosis. Congenital thrombocytopenia, anemia, and petechial rash can be presented immediately after birth.

2- Infantile form is an early-onset, rapidly progressive form that is presented by hepatosplenomegaly, jaundice, hypotonia and neurodevelopmental delay.

3- Juvenile form is a delayed-onset, slowly progressive form that patients are normal in early childhood, and later show mild intellectual impairment, supranuclear vertical gaze paresis, gelastic cataplexy, and ataxia. Dementia and, different types of seizures are appeared.

4- Adult type is a late-onset form, with slowly progressive pattern distinguished by neuropsychiatric disorders.

Managements consist of dietary restriction of cholesterol and administration of Miglastat that are led to a slower deterioration of neurological disorder in these patients.

Keywords: NPC; Children; Mangement

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