Preinatal Types of Niemann-Pick disease type C

Niemann-Pick type C disease is a neurodegenerative atypical lysosomal storage disease. This disease characterized by accumulation of unesterified cholesterol in the lysosomal in the liver, spleen, brain, and lung. Niemann-Pick type C is a autosomal recessive disorder that is rare, progressive, irreversible disease. This disease occurs in approximately: 120,000 to 1/150,000 live births. Niemann-Pick type C has clinical spectrum ranges of clinical presentation from perinatal period to adult-onset chronic neurological and ophthalmological disease. The perinatal presentation is visceral.

Fetal presentation is rare and included in utero splenomegaly or hepatomegaly or ascites and Nonimmune hydrops fetalis and intra uterine growth retardation and oligohydramnios and placentomegaly and fetal death. After birth may be presented with low birth weight and congenital anemia or thrombocytopenia and petechial rash. Some patients died within the first months of life from a rapidly fatal neonatal cholestatic disease. The prenatal ultrasonographic findings of the Niemann-Pick type C include hepatomegaly, ascites and Splenomegaly .each of findings in a fetus, should consider fetal Niemann-Pick type C.

Prolonged cholestatic jaundice in neonate has a great diagnostic value in the Niemann-Pick type C.

The neurological involvement may be occurs after systemic signs such as cholestatic jaundice or hepatosplenomegaly in neonatal period or infancy.

Diagnosis requires skin or fibroblasts culture to confirm accumulation of cholesterol in lysosomes with filipin staining..in some patients molecular genetic testing of Niemann-Pick type C is useful to confirm the diagnosis and essential for prenatal diagnosis.

The differential diagnosis depends on signs and symptomes for example Nonimmune hydrops fetalis or hepatosplenomegaly or cholestatic jaundice in neonats.

Management is symptomatic and crucial.miglustat, is a specific treatment of the neurological manifestations.

Prognosis correlates with the age at onset of the neurological finding.

Keywords: Preinatal; Niemann-Pick type C; Cholestatic jaundice; Hepatosplenomegaly; Lysosomal storage; Filipin staining