

## Approach to NPC Disease

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Neimann-Pick type C is an autosomal recessive disease due to toxic deposition of lysosomal lipids such as glycosphingolipids and unesterified cholesterol causing cell damage. NPC has a wide spectrum of clinical manifestations and time to diagnosis is influenced by the type of symptoms experienced by patients and is varied from a few months to 19 years. Visceral symptoms are the earliest manifestations followed by neurologic signs and psychiatric symptoms which have the longest journey to diagnosis up to adulthood.

Visceral symptoms are presented by hepatosplenomegaly and prolonged neonatal Jaundice, and the patients are usually visited by general pediatrician, metabolic or GI specialist. These clinical presentations are not always recognized in infancy and can subside spontaneously by themselves overtime or attributed to infectious or immune disease.

Neurologic symptoms can manifest as developmental delay, ataxia, gait disturbance, dystonia and reduced academic performance. Supra nuclear vertical gaze palsy and gelastic cataplexy are very specific signs for NPC and appear in late childhood and early adolescence.

Psychiatric symptoms are the latest manifestations. Those symptoms such as behavior and mood problems often emerge in adolescence and are commonly misdiagnosed as autism with psychiatric feature or schizophrania.

**Keywords:** Neimann-pick type C; Approach; Children

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