

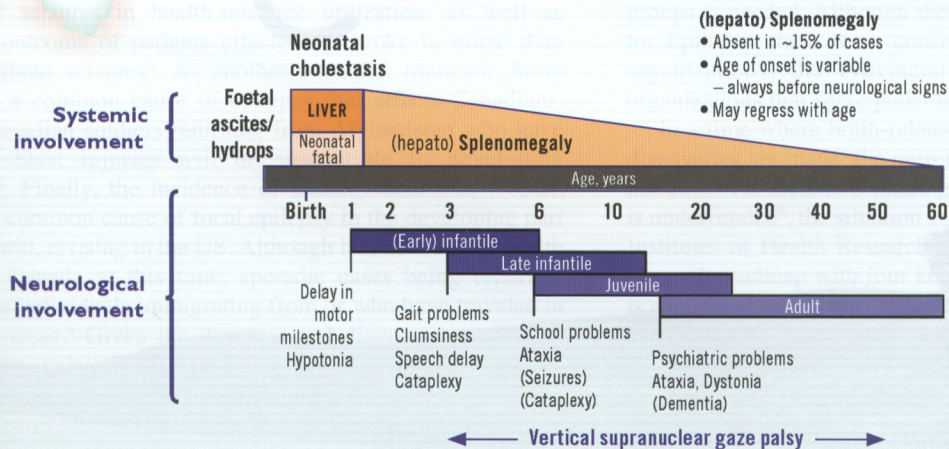
Hepatosplenomegaly +
 Gelastic Cataplexy +
 Dementia +
 Vertical supranuclear Gaze Palsy +
 Ataxia =

Sometimes, x PLUS y equals something surprising.

Be on the lookout for the "PLUS": progressive neurologic manifestations PLUS visceral or psychiatric symptoms. When you see the PLUS, think beyond the individual symptoms. You might be facing **Niemann-Pick Disease Type C (NP-C)**.

- NP-C is a rare inborn error of metabolism affecting children and adults
- NP-C is characterized by a broad clinical variability involving neurological, psychiatric, and visceral symptoms
- NP-C is characterized by progressive neurological symptoms and premature death in most patients

Figure: Time course of selected neurovisceral symptoms of Niemann-Pick disease type C



Early diagnosis of Niemann-Pick Disease Type C is crucial.

When you see the following manifestations:

- Neurologic PLUS...
- Visceral PLUS...
- Psychiatric PLUS...

...consider Niemann-Pick Disease Type C and take action.

Refer patients exhibiting signs and symptoms consistent with NP-C to a metabolic genetics specialist.

For more information: www.npc-si.ca

Reference: 1. Vanier MT. Niemann-Pick disease type C. Orphanet Journal of Rare Diseases 2010; 5:16.



