Niemann-Pick disease type C1

What is Niemann-Pick disease type C1?

It is a rare and incurable progressive disorder sometimes called childhood Alzheimer's because some of its symptoms mirror those of the brain disease.

What causes Niemann-Pick disease type C1?

It is a rare genetic disorder. Patients are not able to metabolize cholesterol and other lipids properly within the cell. Excessive amounts of cholesterol accumulate within the liver and spleen and excessive amounts of other lipids accumulate in the brain.

What are the symptoms of Niemann-Pick type C1?

- Prolonged jaundice can present at birth.
- enlargement of the spleen or liver or enlargement of the spleen or liver frequently becomes less apparent with time
- Progressive neurological disease is the hallmark of Niemann–Pick type C disease, and is responsible for disability and premature death in all cases beyond early childhood.
- Classically, children with NPC may initially present with delays in reaching normal developmental milestones skills before manifesting cognitive decline (dementia).
- Neurological signs and symptoms include :
    - unsteady walking with uncoordinated limb movements
    - slurred speech
    - difficulty in swallowing
    - tremor, epilepsy (both partial and generalized)
    - upgaze palsy, downgaze palsy, saccadic palsy or paralysis
    - sleep inversion,
    - sudden loss of muscle tone or drop attacks

- abnormal movements or postures caused by contraction of agonist and antagonist muscles across joints most commonly begins with in turning of one foot when walking
- ptosis (drooping of the upper eyelid)
- microcephaly (abnormally small head)
- psychosis, progressive dementia
- progressive hearing loss
- Bipolar disorder or major and psychotic depression that can include hallucinations, delusions, mutism

What is the treatment for Niemann-Pick disease type C1?

There is no known cure for this disease but supportive care is essential and can improve the quality of life for people affected with Niemann-Pick disease type C1. The therapeutic team can include specialists in:

- Neurology
- Pulmonology
- Gastroenterology
- Psychiatrist
- Orthopedics
- Nutrition
- Physical Therapy
- Occupational Therapy

What is the prognosis?

The lifespan of patients with NPC is usually related to the age of onset. Children with infantile onset usually succumb in the first few months or years of life, whereas adolescent and adult onset forms of Niemann–Pick type C have a more insidious onset and slower progression, and affected individuals may survive to the seventh decade.

Adult cases of NPC are being recognized with increasing frequency. It is suspected that many patients affected by NPC are undiagnosed, owing to lack of awareness of the disease and the absence of readily available screening or diagnostic tests. For the same reasons the diagnosis is often delayed by many years.

Resources

- **Niemann–Pick disease, type C:** https://en.wikipedia.org/wiki/Niemann%E2%80%93Pick_disease,_type_C
- **Rare Diseases:** https://rarediseases.org/rare-diseases/niemann-pick-disease-type-c
- **The National Niemann-Pick Disease Foundation:** www.nnpdf.org
- **GeneReviews: https://www.ncbi.nlm.nih.gov/books/NBK1296**