Infantile-Onset Niemann-Pick Disease Type C (NPC)
Treatment Landscape

NPC is a rare, fatal genetic disease
caused by *NPC1* or *NPC2* mutations that impair cholesterol trafficking[1,2]
Infantile-onset: neurological symptom onset at ≤6 years (early-onset at ≤2 years and late-onset at 2–6 years)[1]

NPC is characterized by
pathological lysosomal cholesterol accumulation in the brain and peripheral tissues, driving progressive neurodegeneration and premature mortality[1,3]

An unmet need persists for therapies targeting the pathophysiology of NPC
to slow disease progression and improve survival in infantile-onset NPC[4,5]

Current treatment guidelines (2018)
predate recent advances and are being revised; currently they recommend managing NPC with symptomatic supportive therapy[4]

No cure exists for NPC. Current treatment options address neurological symptoms and supportive care only – not the underlying disease pathology[5,6]

Heat shock protein (HSP) activator
HSP70 co-inducer is approved for neurological manifestations of NPC in adults and pediatric patients ≥2 years[8]

Glucosylceramide synthase inhibitor
Substrate reduction therapy is approved in the EU to slow neurological deterioration in adult and pediatric patients with NPC[7]
Not FDA approved for NPC; used off-label in the US

Modified amino acid
N-acetyl amino acid derivative approved for neurological manifestations of NPC in adults and pediatric patients ≥15 kg[9]

Investigational Approaches
Therapeutic approaches targeting NPC pathophysiology mechanisms are being investigated[10,11]

Unmet Need
Despite two FDA approvals, there is no treatment available that addresses the pathophysiology of NPC. Critically, neither is indicated for patients under 2 years of age, leaving earliest-onset and most severely affected patients without a treatment option[5,8,9]

As our understanding of NPC pathophysiology advances, disease management is evolving toward disease-modifying treatment and earlier intervention.

FDA, Food and Drug Administration; EU, European Union; HS70, heat shock protein 70; NPC, Niemann-Pick disease type C; *NPC1*, NPC intracellular cholesterol transporter 1; *NPC2*, NPC intracellular cholesterol transporter 2; US, United States.

1. Berry-Kravis E. *Semin Pediatr Neurol.* 2021;37:100879. 2. Bolton SC, et al. *Orphanet J Rare Dis.* 2022;17(1):51. 3. Winstone AM, et al. *Dev Med Child Neurol*. 2017;59(9):965-972. 4. Geberhiwot T, et al. *Orphanet J Rare Dis.* 2018;13(1):50. 5. Bremova-Ertl T, Patterson M. Niemann-Pick Disease Type C. 2000 Jan 26 [Updated 2025 Nov 20]. Accessed February 26, 2026. https://www.ncbi.nlm.nih.gov/books/NBK1296/. 6. Niemann Pick Disease Type C. National Organization for Rare Disorders. (NORD). Accessed February 26, 2026. https://rarediseases.org/rare-diseases/niemann-pick-disease-type-c/. 7. Zavesca® (miglustat). Summary of Product Characteristics, December 2012. 8. Miplyffa® (arimoclomol). Prescribing Information, October 2025. 9. Aqneursa™ (levacetylleucine). Prescribing Information, September 2024. 10. NCT02534844. ClinicalTrials.gov. Accessed February 26, 2026. https://clinicaltrials.gov/study/ NCT02534844. 11. NCT04860960. ClinicalTrials.gov. Accessed February 26, 2026. https://clinicaltrials.gov/study/NCT04860960.

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