NPC GenomeComplete Sponsored Testing Program

Beren Therapeutics P.B.C., in partnership with GeneDx, is offering a no-cost whole genome testing program for individuals with suspected Niemann-Pick disease type C (NPC), an ultra-rare, underdiagnosed, progressive genetic disorder that affects the body's ability to transport cholesterol and other lipids within cells.

Is your patient eligible? Patients must meet all of the following criteria:

- ✓ **Reside in the United States**
- ✓ **Must be age 15 or younger**
- ✓ **Parent/Guardian must consent to sharing de-identified data with Beren Therapeutics P.B.C.**

And ONE of the following:

- Clinical Findings – 3 or more of the following
  - Supranuclear gaze palsy or supranuclear saccadic palsy. *Inability to voluntarily move eyes up or down.*
  - Developmental regression, early-onset cognitive decline, or dementia
  - Dysarthria or dysphagia
  - Treatment-resistant psychiatric illness (psychosis, schizophrenia, depression, etc.)
  - Acute neonatal liver failure
  - Pulmonary infiltrates or respiratory failure in infancy
  - Splenomegaly and/or hepatomegaly
  - Gelastic cataplexy. *Sudden, brief loss of muscle tone triggered by laughter*
  - Progressive ataxia, dystonia, spasticity, or cerebral palsy
  - Seizures
  - Sensorineural hearing loss
  - Prolonged, unexplained jaundice or cholestasis (>2 weeks)
  - Hypotonia or failure to thrive
  - Unexplained fetal ascites or non-immune hydrops fetalis
- Family history of Niemann-Pick disease type C with a confirmed molecular diagnosis
- Elevated NPC Biomarker
  - C-triol, PPCS, PPCS:lyso-SM, or bile acid derivative (plasma or urine)

Your patient's data stays private

GeneDx will never share your patient's protected health information, personally identifiable information, or raw sequencing data with any third party.

How to order

1. Login or sign up for a GeneDx account and add GenomeDx® or GenomeDx® Rapid, (proband, duo or trio) or Targeted Variant Testing for *NPC1* or *NPC2* to your cart
2. Select the program code **STP-BEREN-NPC**
3. Confirm your patient's eligibility
4. Follow the prompts and enter in the appropriate information
5. Place your order and GeneDx will follow up when your results are ready

Scan the QR code to get started

For access to the program-specific test requistion form, please scan the QR code or visit **genedx.co/beren-npc**

Find answers with whole genome sequencing

Whole genome sequencing provides the most complete picture of an individual's DNA, detecting variants across coding and non-coding regions as well as structural changes missed by other methods. This comprehensive view increases the chance of finding meaningful answers in complex, atypical, or undiagnosed cases. GenomeDx™ clinical genome sequencing is performed at ≥40× average coverage across the genome (minimum 30×), with ~97% of coding regions assessed at ≥15× coverage. Analysis includes SNVs, CNVs >1 kb, select repeat expansions, SMN1 exon 8 loss, and uniparental disomy (when parents are submitted). All testing is performed in GeneDx's CLIA-certified and CAP-accredited laboratory. To learn more, see our GenomeDx® test information sheet.

Remove barriers to testing. Empower earlier insights.

Images are illustrative only and not actual patients

GeneDx® beren™