

NPC GenomeComplete Sponsored Testing Program

Beren Therapeutics P.B.C., in partnership with GeneDx, is offering a no-cost genetic 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.



Talk to your doctor about whether this program may be right for your loved ones.

Is your child 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. *Slurred or difficult to understand speech or trouble swallowing*
 - Treatment-resistant psychiatric illness (psychosis, schizophrenia, depression, etc.)
 - Acute neonatal liver failure
 - Pulmonary infiltrates or respiratory failure in infancy
 - Splenomegaly and/or hepatomegaly. Enlarged spleen and/or liver
 - Gelastic cataplexy. *Sudden, brief loss of muscle tone triggered by laughter*
 - Progressive ataxia, dystonia, spasticity, or cerebral palsy. *Worsening movement disorder*
 - Seizures
 - Sensorineural hearing loss
 - Prolonged, unexplained jaundice or cholestasis (>2 weeks)
 - Hypotonia or failure to thrive
 - Unexplained fetal ascites or non-immune hydrops fetalis. *Unexplained fluid buildup in the baby's belly or body during pregnancy*
- Family history of NPC
- Elevated NPC Biomarker
 - C-triol, PPCS, PPCS:lyso-SM, or bile acid derivative (plasma or urine)

Your data stays private

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

To order a test for your patient, follow these steps:

Only licensed medical professionals can order a test.

- 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 NPC 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 requisition form, please scan the QR code or visit genedx.com/providers/genetic-testing-programs/beren-npc

Find answers with genome sequencing

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 $\geq 40\times$ average coverage across the genome (minimum $30\times$), with $\sim 97\%$ of coding regions assessed at $\geq 15\times$ 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.



Remove barriers to testing.
***Empower* earlier insights.**

Images are illustrative only and not actual patients

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