PERSEVERE

Persist. Quest. Cure.

PERSEVERE – To persist in an undertaking in spite of counter-influences, oppositions or discouragements.

"We, the families of the children and adults affected by Niemann-Pick Disease,

Thank You

for joining us as we persevere in our quest to find a cure."

National Niemann-Pick Disease Foundation
P.O. Box 310
Fort Atkinson, WI 53538-0310

Tele: 920-563-0930 • Email: nnpdf@nnpdf.org

www.nnpdf.org

The National Niemann-Pick Disease Foundation

The National Niemann-Pick Disease Foundation (NNPDF) was established in 1992. It has grown to become an international, voluntary, non-profit organization comprised of parents, relatives and friends committed to finding a cure for all types of Niemann-Pick Disease.

Our primary goals:

- Promote research into the causes of Niemann-Pick Disease (NPD)
- Provide information to assist in the correct medical diagnoses and referrals of children with NPD
- Facilitate genetic counseling for parents who are known NPD carriers
- Encourage the exchange of research findings among scientists
- Support legislation that positively impacts patients and families affected by NPD

While there is little that can ease the emotional burden of NPD, interaction with other parents and families reduces feelings of isolation and despair.

That's why the National Niemann-Pick Disease Foundation strives to:

- Give and facilitate emotional support
- Provide assistance during a crisis
- Share resources and ideas including, but not limited to, doctors, clinics, insurance companies and additional health and human services
- Provide practical suggestions about the day-to-day care of those with NPD
- Establish enduring relationships with other families affected by NPD

The vision of the members of the NNPDF is that individuals affected by Niemann-Pick Disease will have the same chance as their siblings and peers to run and play, to hope and achieve, and to live out their dreams.

Please fill out the attached donation form and mail with your tax-deductible contribution to:

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Research Is Our Only Hope

Researchers are working to improve methods of diagnosing and treating Niemann-Pick Disease Type C (NPC). Developing accurate, early diagnostic tools and life-saving treatments for NPC are complex endeavors, but progress is being made. Investigations into the basic cellular processes involved in cholesterol metabolism have led to discovery of the two genes responsible for NPC. Initial studies of these genes' roles in the cell have helped identify several potential disease-modifying therapies. Other treatments may be on the horizon as more is learned about the disease process. Current disease management focuses on improving quality of life through symptomatic therapy, but much work remains to be done!

Research is the key to finding effective treatments, and one day, a cure for NPD. We will PERSEVERE until the battle against NPD is won! We need your help to achieve this goal.

They have only their childhood to *experience a lifetime.*

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The horror of Niemann-Pick Disease Type C (NPC) is too great for most of us to imagine.

This rare, deadly disease robs a child of their most precious gift - life. When a family receives the soul-crushing diagnosis of NPC for their child or loved one, an incomprehensible journey of gradual decline, suffering and death is a certainty.

Researchers are working diligently to learn as much as they can about NPC, but this is an uphill battle. NPC is highly unpredictable and the symptoms are similar to dozens of other conditions. Significant progress has been made, but diagnosis is still difficult and a treatment has yet to be found.

You can help children and families facing NPC. Please, take a few moments to read and learn about the disease. Further, we ask you to reach out and offer support to families affected by Niemann-Pick Disease. They are struggling to face the challenges of this devastating disease and the eventual loss of a child or loved one, and they desperately need the support of family, friends, and their communities.

Please consider making a tax-deductible donation to the NNPDF's programs of research and family support services. We are working toward a day when NPC can be quickly and accurately diagnosed and treated with 100 percent success. We are looking forward to the day when we can unite in celebration and shout, "We have prevailed in our Quest for a Cure!" Until then, we will PERSEVERE in the battle against this devastating disease. Together, we can make it happen. Together, we can make a difference!

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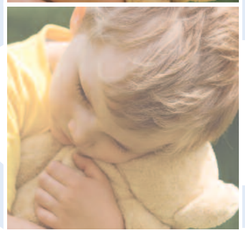


The complexities of Niemann-Pick Disease Type C

The term "Niemann-Pick Disease" (NPD) refers to two categories of disease: 1) NPD due to Acid Sphingomyelinase Deficiency (ASMD), also known as NPD Types A, A/B, and B, and 2) NPD Type C (NPC), characterized by abnormal cholesterol and lipid processing in the cell, and caused by mutations in either of two genes, *NPC1* or *NPC2*.

(For more information about Niemann-Pick Disease Types A and B, also known as Acid Sphingomyelinase Deficiency, or ASMD, please see the NNPDF's companion brochure.)

NPC occurs in all ethnic groups. It is inherited as an autosomal recessive condition. This means that affected individuals have two altered copies of either *NPC1* or *NPC2*, having inherited one copy from each parent. Each unaffected parent (called a carrier) of an affected individual has one altered copy of the disease-causing gene and one normally-functioning copy of that gene. For a couple who are both carriers, there is a 1 in 4 chance with each pregnancy that a child will be affected by NPC, a 2 in 4 chance that the child will be a carrier of NPC, like the parents, and a 1 in 4 chance that the child will be neither a carrier nor affected.

NPC is an exceptionally variable condition. The symptoms of the disease are shared by a number of related lysosomal disorders. Further, the rate of disease progression varies from patient to patient, even within families where more than one child is affected. This variability makes it extremely difficult to recognize and diagnose NPC, and often leads to delay in confirmation of the diagnosis.

NPC is often classified into three groups, recognizing that, in reality, there is considerable overlap and that every patient is unique:

1) Early-onset NPC (early and late infantile)
This form of the disease can begin before delivery, at delivery, or throughout the first two years of life.
Symptoms can include:
a. Abnormal build-up of fluid in the abdomen, called ascites
b. Enlarged liver and spleen
c. Severe liver disease with jaundice that can last for weeks or months
d. Respiratory failure
e. Very early death in some cases
f. Decreased muscle tone (hypotonia)
g. Developmental delay

2) Classic childhood onset NPC
This form of the disease is often first recognized at about the time a child is entering school. Signs may be subtle at first, but gradually become more noticeable, and can include:
a. Clumsiness, unsteadiness of gait, problems walking
b. Enlarged liver and/or spleen – but this may be absent or appear to resolve
c. Difficulty with voluntary upward and downward eye movements
d. Learning difficulties and worsening intellectual impairment (with eventual dementia)
e. Slurred speech and swallowing difficulties
f. Sudden loss of muscle tone, leading to falls (cataplexy)
g. Seizures
h. Sleep disturbance

3) Adult or late-onset NPC
Occurring from later adolescence into and through adulthood, this variant may include:
a. Neurologic disease as seen in childhood onset NPC
b. Major psychiatric illness (schizophrenia, depression, psychosis) with subtle, often unrecognized neurologic signs
c. Dementia

Current treatment strategies for NPC focus on management of symptoms to improve the quality of life for affected individuals and their families. These include, but are not limited to:
a. Control of seizures, cataplexy and other neurologic symptoms
b. Evaluation and treatment of sleep difficulties
c. Chest physical therapy and prevention of pneumonia
d. Physical therapy to maintain mobility
e. Swallowing and nutrition assessment; G-tube placement
f. Routine developmental and skills testing

National Niemann-Pick Disease Foundation

DONATION FORM

Please help us in our fight against Niemann-Pick Disease. Your tax-deductible contribution helps to support our programs of research for Niemann-Pick Disease, support our NPD families and educate the public.

Thank You
for joining us as we persevere in our quest for a cure.

Name: _____

Organization/Business: _____

_____

Address: _____

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In Honor of/In Memory of: _____

_____

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The National Niemann-Pick Disease Foundation has been designated a 501 (c) (3) organization and contributions are tax-deductible to the extent provided by law.