

Dear Doctor,

I have Niemann-Pick Disease Type C. Niemann-Pick Disease type C (NPC) is caused by mutations in either the NPC1 and NPC2 genes that result in impaired cellular processing and transport of low-density lipoprotein (LDL) cholesterol. The broad clinical spectrum ranges from a rapidly fatal neonatal disorder to an adult-onset chronic neurodegenerative/psychiatric disease. The neurological involvement defines the disease severity in most patients but is typically preceded by systemic signs (cholestatic jaundice in the neonatal period or isolated splenomegaly or hepatosplenomegaly in infancy or childhood). The first neurological symptoms vary with age of onset: delay in developmental motor milestones (early infantile period), gait problems, falls, clumsiness, cataplexy, school problems (late infantile and juvenile period), and ataxia that may be noted following initial psychiatric disturbances (adult form). The most characteristic sign is vertical supranuclear gaze palsy. The neurological disorder consists mainly of cerebellar ataxia, dysarthria, dysphagia, and progressive dementia. Cataplexy, seizures and dystonia are other common features (Vanier 2010). The website listed below provides a link to the full text of Dr. Vanier's landmark paper.

NPC has a wide range of symptoms and severity. Listed below are some of the common issues people with NPC deal with. In the past, I have had the following symptoms of NPC:

- | | |
|-------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------|
| <input type="checkbox"/> clumsiness and gait abnormalities | <input type="checkbox"/> dysarthria and dysphagia (difficulty with speech and swallowing) |
| <input type="checkbox"/> ataxia (severe clumsiness) | <input type="checkbox"/> history of aspiration |
| <input type="checkbox"/> cognitive impairment | <input type="checkbox"/> seizure |
| <input type="checkbox"/> vertical supranuclear ophthalmoplegia (inability to look up/down) | <input type="checkbox"/> hepatic and splenic disease from infiltration |
| <input type="checkbox"/> gelastic cataplexy (laughing or smiling and then losing muscle control briefly) | <input type="checkbox"/> abnormal blood counts ie: platelets and red blood cells/anemia/thrombocytopenia |
| <input type="checkbox"/> hearing impairment | <input type="checkbox"/> pulmonary disease secondary to alveolar proteinosis |
| <input type="checkbox"/> constipation | <input type="checkbox"/> psychiatric symptoms |
| <input type="checkbox"/> dystonia (muscle weakness) | |

For a complete review on NPC, please see Dr. Marie Vanier's review from the *Orphanet Journal of Rare Diseases* at nnpdf.org/files/2018/07/Vanier-NPC-10.2009.pdf

Consensus Clinical Management Guidelines published in 2018 can be seen at nnpdf.org/files/2018/07/CCM-Guidelines-2018.pdf

My Health Now

Date: _____

The reason for this emergency...

The specific health issues bothering me **RIGHT NOW** and the date they started are listed here:

Current treatments or medications for this issue:

I am/am not on a special or restricted diet. Dietary restrictions are listed here:

I am under medical care for this current issue. My managing physician is:

Name: _____

Clinic or Hospital: _____

Phone/Contact Info: _____

IMPORTANT: In addition to the information you keep in this booklet, please keep the following items with you:

- Identification and insurance cards
- Please bring a copy of recent lab work and imaging studies if applicable

Basic Information

Name: _____

Address: _____

City, State, Zip: _____

Date of Birth: _____ Height: _____ Weight: _____

Blood Type: _____ SSN: _____

Primary Care Physician: _____

Hospital/Clinic: _____

Phone/Contact Info: _____

NPC Specialist: _____

Hospital/Clinic: _____

Phone/Contact Info: _____

Specialist: _____

Hospital/Clinic: _____

Phone/Contact Info: _____

Specialist: _____

Hospital/Clinic: _____

Phone/Contact Info: _____

Specialist: _____

Hospital/Clinic: _____

Phone/Contact Info: _____

Health Insurance:

Company: _____

Policy Number: _____

Phone/Contact Info: _____

In Case of an Emergency

Primary emergency contact person:

Name: _____ Relationship: _____

Address: _____

City, State, Zip: _____

Phone/Contact Info: _____

Alternate emergency contact person:

Name: _____ Relationship: _____

Address: _____

City, State, Zip: _____

Phone/Contact Info: _____

Who can make medical decisions for me:

Name: _____ Relationship: _____

Address: _____

City, State, Zip: _____

Phone/Contact Info: _____

Living Will/Advanced Directive previously completed: Yes No

If yes, please bring a copy.

Organ Donor: Yes No

Please provide copy of Health Care Proxy if available

I am currently or have recently participated in a clinical trial for NPC: Yes No

If yes, please provide the following:

Investigator Name & Phone/Contact Info: _____

Institution Name: _____

Treatment Studied: _____

My three most active health issues are:

| |
|----|
| 1. |
| 2. |
| 3. |

Medications and prescribed treatments that I am currently taking including dosage and frequency:

| Medication & Treatments | Dosage & Frequency |
|-------------------------|--------------------|
| | |
| | |
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Medications for other conditions that I am currently taking including dosage and frequency:

| Medication | Dosage & Frequency |
|------------|--------------------|
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| | |

My Current Pharmacy:

Name: _____

City and State: _____

Phone/Contact Info: _____

Supplements, vitamins, and alternative medications including dosing and frequency:

| Supplements, Vitamins, etc. | Dosage & Frequency |
|-----------------------------|--------------------|
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Allergies

Allergies to medications including reactions I had: _____

Other allergies:

My dietary restrictions include:

If I use supplemental feeding, I currently take:

Major Illnesses/Chronic Diseases

| Major Illness/Chronic Disease | Date Occurred/ Diagnosed | Treatment |
|-------------------------------|-----------------------------|-----------|
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Hospitalizations

| Date | Reason | Treatment |
|------|--------|-----------|
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Surgeries/Procedures

| Date | Surgery/Procedure | Result |
|------|-------------------|--------|
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Physical Assistance Hearing aids, eye glasses, mobility aids

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Immunizations

| Immunization | Date |
|------------------------------------------------------------|------|
| Diphtheria, Tetanus, and Pertussis (DTaP) | |
| Tetanus, Diphtheria (Td) | |
| Influenza (Flu) | |
| Hepatitis B (List number/date of last 3 part series given) | |
| Hepatitis A (List number/date of last 2 part series given) | |
| Measles, Mumps, & Rubella (MMR) | |
| Chickenpox | |
| Shingles (Also called Herpes Zoster or Zoster) | |
| Haemophilus Influence Type b (Hib) | |
| Rotavirus | |
| Pneumococcal Conjugate (PCV13) (Pneumonia) | |
| Polio (4 doses of IPV for kids - List date of #4) | |
| Meningococcal (Meningitis) | |
| Human Papillomavirus - Gardasil® (HPV) | |
| TB Screen | |
| | |
| | |
| | |

Family History

Include major conditions such as diabetes, heart attacks, stroke, kidney failure, cancer, COPD/asthma, etc.

| Relative | Major Condition or Ailment | Age or Year and Age at Death | Caused /Contributed to death? |
|----------|----------------------------|------------------------------|----------------------------------------------------------|
| | | | <input type="checkbox"/> Yes <input type="checkbox"/> No |
| | | | <input type="checkbox"/> Yes <input type="checkbox"/> No |
| | | | <input type="checkbox"/> Yes <input type="checkbox"/> No |
| | | | <input type="checkbox"/> Yes <input type="checkbox"/> No |
| | | | <input type="checkbox"/> Yes <input type="checkbox"/> No |
| | | | <input type="checkbox"/> Yes <input type="checkbox"/> No |
| | | | <input type="checkbox"/> Yes <input type="checkbox"/> No |
| | | | <input type="checkbox"/> Yes <input type="checkbox"/> No |

About the NNPDF

The National Niemann-Pick Disease Foundation, Inc., a 501(c) (3) membership organization is the U.S. unbiased family service and advocacy group dedicated to supporting and empowering patients and families affected by Niemann-Pick disease through education, collaboration and research. We envision an enduring patient support and advocacy community that embodies strength, hope and empathy for those affected by all Niemann-Pick Disease types. We believe deeply in the future of research that promotes clinical treatments with potential to improve patient quality of life and with intent to ultimately cure all Niemann-Pick Disease types.

We are committed to:

- Supporting and empowering families, caregivers and patients from diagnosis, through disease progression, including therapies and grief.
- Providing information and referral for health care issues.
- Managing a network of family members who can provide support and information to fellow patients/families throughout the journey.
- Partnering within industry representatives considering or currently investing in therapeutic treatments for Niemann-Pick disease.
- Overseeing community representation and education regarding drug development and regulatory process.

To learn more about NNPDF and its resources: please visit us at nnpdf.org.

To support NNPDF: Charitable donations to support our programs and services are always appreciated. To make a gift online please visit: nnpdf.org/fundraising/make-a-donation/

Or you can mail your gift to:

NNPDF | P.O. Box 49 | Fort Atkinson, WI 53538-0049

Please make checks payable to:

National Niemann-Pick Disease Foundation or NNPDF

The NNPDF designed this document to assist health care providers in the care they provide individuals with NPC. We hope this tool will allow families to provide consistent and complete information at all medical encounters. If you have thoughts or suggestions on how we can help including thoughts on this document, please provide feedback to us at nnpdf@nnpdf.org.

THANK YOU!

Acknowledgement

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