



# My Health Handbook & Emergency Information

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I have Niemann-Pick Disease Type C

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This booklet contains information you should know about me in an emergency

PROVIDED BY



NATIONAL NIEMANN-PICK DISEASE FOUNDATION, INC.  
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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<br>(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<br>(inability to look up/down)               | <input type="checkbox"/> hepatic and splenic disease from infiltration                                      |
| <input type="checkbox"/> gelastic cataplexy (laughing or smiling and<br>then losing muscle control briefly) | <input type="checkbox"/> abnormal blood counts ie: platelets and red<br>blood cells/anemia/thrombocytopenia |
| <input type="checkbox"/> hearing impairment   | <input type="checkbox"/> pulmonary disease secondary to alveolar<br>proteinosis                             |
| <input type="checkbox"/> constipation   | <input type="checkbox"/> psychiatric symptoms   |
| <input type="checkbox"/> dystonia (muscle weakness)   |   |

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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](http://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](http://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

**Medications for other conditions that I am currently taking including dosage and frequency:**

Medication	Dosage & Frequency

**My Current Pharmacy:**

Name: \_\_\_\_\_

City and State: \_\_\_\_\_

Phone/Contact Info: \_\_\_\_\_

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

Supplements, Vitamins, etc.	Dosage & Frequency

## 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

## Hospitalizations

Date	Reason	Treatment

## Surgeries/Procedures

Date	Surgery/Procedure	Result

## Physical Assistance Hearing aids, eye glasses, mobility aids


## 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](http://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/](http://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](mailto:nnpdf@nnpdf.org).

## THANK YOU!

### **Acknowledgement**

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