

Evaluating Niemann-Pick disease type C (NPC) symptoms

How to use this guide

Identifying symptoms is important for a more timely and accurate diagnosis of Niemann-Pick disease type C (NPC). With a diagnosis, you can start managing the symptoms of NPC, and potentially address the underlying disease. This guide is intended to help you or your loved one **start the conversation with your doctor** about a suspected NPC diagnosis. Below is a checklist of symptoms and some questions to help guide discussion.

NPC is a genetic disease, so you may want to consider genetic testing for you and your loved ones.

Share LearnNPC.com with your doctor and explore Invitae.com, the genetic testing site, together.

Testing is readily available, may be free of charge for those who qualify, and includes genetic counseling.

NPC symptoms checklist

NPC can present with a wide range of symptoms that may begin in infancy. Symptoms can vary from person to person, may be more severe for some, and can change as the person gets older.

Review the questions below and check all responses that apply to you or your loved one.

Are you or a loved one experiencing symptoms of NPC?

- | | |
|---|---|
| <input type="radio"/> Enlarged liver or spleen (hepatosplenomegaly) | <input type="radio"/> Learning difficulties |
| <input type="radio"/> History of jaundice | <input type="radio"/> Abnormal muscle contractions (dystonia) |
| <input type="radio"/> History of seizures | <input type="radio"/> Problems sleeping |
| <input type="radio"/> Low muscle tone | <input type="radio"/> Difficulty swallowing |
| <input type="radio"/> Developmental delays | <input type="radio"/> Slow or slurred speech |
| <input type="radio"/> Difficulty with eye movements* | <input type="radio"/> Psychiatric illness such as psychosis or depression |
| <input type="radio"/> Unsteadiness or frequent falls | <input type="radio"/> Behavioral problems |
| <input type="radio"/> Problems with coordination† | <input type="radio"/> Cognitive impairment including dementia |
| <input type="radio"/> Abnormal posture | |

*Tracking with the eyes, particularly looking up or down.

†Including muscle spasms that impact ability to use hands to hold objects, write, eat independently.

NPC is a neurodegenerative and progressive disease. The age at which symptoms start can vary from person to person, and the way they manifest can look different.

At what age did you or a loved one first notice symptoms?

- | | | |
|--|--------------------------------------|-------------------------------------|
| <input type="radio"/> <2 months old | <input type="radio"/> 2–6 years old | <input type="radio"/> >15 years old |
| <input type="radio"/> 2 months–2 years old | <input type="radio"/> 6–15 years old | |

The severity of symptoms may change over time depending on the age at which they started.

NPC is a rare disease and may be misdiagnosed. Have you or your loved one been diagnosed with any of the following conditions?

- ☐ Dyspraxia
- ☐ Cerebral palsy
- ☐ Parkinson's disease
- ☐ Psychiatric disorders
- ☐ Other

How are these symptoms affecting your day-to-day life?

Questions to ask your doctor

"What tests can we do to confirm or rule out NPC or similar conditions?"

"Based on the symptoms we've documented, do you think Niemann-Pick disease type C could be a possibility?"

"Are there specific symptoms or changes we should watch closely and report right away?"

"What treatment options are available and may be appropriate for my NPC?"

Additional questions:

Start the conversation with your doctor today.

Recognize NPC symptoms and work with your doctor to get a diagnosis as soon as possible.



Visit LearnNPC.com to learn more about NPC and the importance of genetic testing.