

**THINK AGAIN
THINK NP-C**

Talk NP-C



Ataxia

is a key symptom of Niemann-Pick type C disease

Patients with **Niemann-Pick type C (NP-C)** may present with progressive ataxia. This may manifest as poorly coordinated movements affecting walking, speech or manipulation.¹ Ataxia may start at any age from late childhood to adulthood.¹

How you might hear ataxia described...

Listen out!

- ◆ He has difficulty walking
- ◆ I fall over a lot
- ◆ I have problems walking in a straight line
- ◆ People ask me if I'm drunk because I'm unbalanced
- ◆ She wobbles when she walks
- ◆ She can't ride her bike anymore
- ◆ He used to love playing football but he can't anymore as he can't run

Patient Insight

“ We noticed that Ben used to bump into things a lot when he was walking – he sometimes wobbled on his feet and walked with his feet quite far apart.

It's most upsetting for Ben when passers-by think he is drunk because of his gait. ”

Healthcare Professional Insight

“ Ataxia is relatively simple to diagnose, however, it may not be apparent during the early stages of the disease. A key indicator of ataxia is the inability to balance.

When assessing gait, patients should be asked to walk in a straight line, turn and also balance on one leg. The inability to make these controlled movements would strongly suggest ataxia. ”

What is Niemann-Pick Type C Disease?

Niemann-Pick type C disease (NP-C) is a rare, progressive, irreversible and chronically debilitating lysosomal storage disease² with an incidence of approximately 1 in 90,000 live births.³ It is an inherited condition and can present at any age, affecting infants, children, adolescents and adults.

NP-C is commonly undetected or misdiagnosed. This is often due to its highly variable clinical presentation, characterised by a wide range of symptoms like ataxia, that individually, are not specific to the disease.^{1,4,5}

References

1. Patterson M, Hendriksz, Walterfang M, *et al.* on behalf of the NP-C Guidelines Working Group. Recommendations for the diagnosis and management of Niemann-Pick disease type C: an update. *Mol Genet Metab* 2012; **106**(3): 330–344.
2. Vanier, M. Niemann-Pick disease type C. *Orphanet J Rare Dis* 2010; **5**: 16.
3. Wassif C, Cross J, Iben J, *et al.* High incidence of unrecognized visceral/neurological late-onset Niemann-Pick disease, type C1, predicted by analysis of massively parallel sequencing data sets. *Genet Med* 2016; **18**(1): 41–48.
4. Wijburg FA, Sedel F, Pineda M, *et al.* Development of a suspicion index to aid diagnosis of Niemann-Pick disease type C. *Neurology* 2012; **78**(20): 1560–1567.
5. Mengel E, Künnemann H, Lourenço C, *et al.* Niemann-Pick disease type C symptomatology: an expert-based clinical description. *Orphanet J Rare Dis* 2013; **8**: 166.

For more information about where to refer patients suspected of having NP-C go to www.think-npc.com