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 have problems walking in a straight line
- She wobbles when she walks
- I fall over a lot
- People ask me if I'm drunk because I'm unbalanced
- 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 disease[2] 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

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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, Klünemann 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

This is a project co-ordinated by the International Niemann-Pick Disease Alliance

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