

**THINK AGAIN  
THINK NP-C**

Talk NP-C

## Dysarthria

*is a key symptom of Niemann-Pick type C disease*

Dysarthria is characterised by irregular and slurred speech arising from an inability to control the muscles of the mouth. Patients with dysarthria often have difficulty controlling the volume and pitch of their speech.<sup>1</sup> Dysarthria results from a combination of symptoms that are commonly seen in **Niemann-Pick type C disease (NP-C)**: ataxia (loss of voluntary muscle control) and dystonia (abnormal muscle tone resulting in muscle spasms).<sup>2-5</sup>

### How you might hear dysarthria described...

Listen out!

- ◆ She finds it hard to speak loudly
- ◆ He talks very slowly
- ◆ It sounds like she speaks through her nose
- ◆ He can't control his pitch
- ◆ He sounds like he's drunk
- ◆ I often can't understand what she's saying
- ◆ He suddenly speaks loudly for no reason
- ◆ She makes gurgly sounds
- ◆ It looks like he finds it hard to speak

### Patient Insight

“ We couldn't understand why Amelia was struggling to speak properly; when she was six years old, her teachers started to comment on Amelia mumbling her words at school. At first we thought she was just playing, as children do, when she would shout and whisper in the same sentence. As time went on Amelia began to slur some of her words. At that point we were referred by our general practitioner to a neurologist who confirmed Amelia had dysarthria. ”

### Healthcare Professional Insight

“ Dysarthria presents in patients as slurred, monotone speech due to difficulty in moving the tongue and muscles around the mouth. Patients will often speak in short sentences and give one word answers. Dysarthria is commonly associated with dysphagia (another symptom of NP-C), which causes difficulty swallowing; therefore, patients may also drool when speaking. ”

### 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<sup>4</sup> with an incidence of approximately 1 in 90,000 live births.<sup>6</sup> 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 dysarthria, that individually, are not specific to the disease.<sup>2,4,6</sup>

#### References

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2. 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.
3. 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.
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5. 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.
6. 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.

For more information about where to refer patients suspected of having NP-C go to [www.think-npc.com](http://www.think-npc.com)