THINK AGAIN
THINK NP-C

Talk NP-C

Gelastic Cataplexy

is a key symptom of Niemann-Pick type C disease

Patients with **Niemann-Pick type C (NP-C)** may present with gelastic cataplexy. Gelastic cataplexy is characterised by a sudden loss of muscle tone (without loss of consciousness), which is typically triggered by an emotional stimulus, for example laughter. Loss of tone may involve the legs, neck or jaw. Therefore, cataplexy may manifest as sudden falls, sudden head drop or jaw drop, triggered by laughing.

How you might hear cataplexy described...

Listen out!

- She laughs so much that she falls down
- I experience drop attacks
- I suddenly lose control of my muscles
- He nods his head a lot
- He loses voluntary control of his limbs
- She seems to faint from emotion
- I collapse suddenly

Patient Insight

"Nina was seven when we were told that she suffers from gelastic cataplexy. We knew that something was wrong after she collapsed at school on three separate occasions. Nina's teacher said that she was just playing with her friends; thankfully, after she fainted, Nina would recover really quickly and luckily she was never seriously injured.

After Nina was diagnosed, she had to make a conscious effort not to laugh or get too angry. It's difficult to leave Nina by herself because we worry that she might collapse and seriously hurt herself."

Healthcare Professional Insight

"Gelastic cataplexy can often be confused with seizures, however, unlike seizures, cataplexy only occurs during periods of emotional stimulation. Once this stimulation has worn off, the person will regain control. Cataplexy can be controlled with medication and protective headgear is available."

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[1] with an incidence of approximately 1 in 90,000 live births.[2] 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 gelastic cataplexy, that individually, are not specific to the disease.[3–5]

References

1. Vanier, M. Niemann-Pick disease type C. *Orphanet J Rare Dis* 2010; **5**: 16.
2. 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.
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.
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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