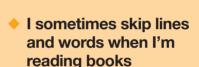
Vertical Supranuclear Gaze Palsy (VSGP)

is a key symptom of Niemann-Pick type C disease

Vertical supranuclear gaze palsy (VSGP) is an eye-movement disorder characterised by difficulty with spontaneous up and down eye movements, along with saccadic palsy or paralysis.¹ VSGP is a typical symptom of **Niemann-Pick type C disease (NP-C)** and is present in virtually all patients with NP-C.^{1,2,4} It often goes undetected, despite it being simple to identify by testing for impairment of voluntary saccades, which is an early sign of NP-C.^{1,2,3}

How you might hear VSGP described...

- She can't focus her eyes
- He struggles to move his eyes quickly and spontaneously
- He has glazed eyes
- ♦ I'm slow to look at things
- She can't see where I'm pointing
- He moves his head to look around rather than his eyes





Patient Insight

When James was 12 we noticed that he began to take extra care when walking up the stairs. Instead of moving his eyes up and down to navigate his way up the staircase he would need to move his whole head.

By the age of 13 it was more noticeable. When James was playing football he would tilt his head to see where his team mates were rather than moving his eyes. We knew that something wasn't quite right.

Healthcare Professional Insight

VSGP is relatively simple to detect. Patients will display distinct eye movement abnormalities, for example, difficulty in fast eye movements between two fixed points (saccadic eye movements) and following moving stimulus within their field of vision (pursuit eye movements). VSGP may be missed if voluntary saccades are not assessed. In NP-C vertical saccadic eye movements are often the first to be affected.

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 VSGP, that individually, are not specific to the disease.^{1,4,5}

References

- 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.
- Wassif C, Cross J, Iben J, et al. High incidence of unrecognized viscoeral/neurological late-onset Niemann-Pick disease, type C1, predicted by analysis of massively parallel sequencing data sets. Genet Med 2016; 18(1): 41–48.
- 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.
- 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

