

**THINK AGAIN  
THINK NP-C**

Talk NP-C

## Hepatosplenomegaly

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

Hepatosplenomegaly is present in almost half of all patients diagnosed with **Niemann-Pick type C disease (NP-C)**.<sup>1</sup> The intensity varies between patients, but is characterised by an enlarged liver and spleen and typically presents as a distended abdomen in younger patients and neonates.<sup>1</sup> It is more difficult to recognise in adults as it is usually asymptomatic, requiring an abdominal ultrasound to diagnose the symptom in suspected cases.<sup>2</sup>

### How you might hear hepatosplenomegaly described...

- ◆ He has a swollen tummy
- ◆ I often feel nauseous
- ◆ He complains of pain in his right side
- ◆ He often has a stomach ache
- ◆ She complains of feeling full a lot
- ◆ Her skin is yellow in colour

Listen out!

### Patient Insight

“Chloe was born with a large, swollen tummy. After a few days her skin began turning pale yellow in colour, which we were told was a sign of jaundice and can be quite common in new born babies. Chloe’s tummy remained large and swollen months after we left the hospital and she was a very sickly baby; vomiting all the time. We were worried. It was only after a liver function test that we were told her jaundice resulted from hepatosplenomegaly.”

### Healthcare Professional Insight

“Hepatosplenomegaly is most commonly detected in younger patients due to the enlarged liver and spleen being more prominent as a swelling in the abdomen. This symptom can be seen in older patients. However, in adults, particularly if they’re overweight, it is more difficult to detect. Jaundice, fatigue and nausea are all symptoms consistent with hepatosplenomegaly.”

### 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>1</sup> with an incidence of approximately 1 in 90,000 live births.<sup>3</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 hepatosplenomegaly that, individually, are not specific to the disease.<sup>2,4,5</sup>

#### References

1. Vanier, M. Niemann-Pick disease type C. *Orphanet J Rare Dis* 2010; **5**: 16.
2. 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.
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](http://www.think-npc.com)