

IN FOCUS

NIEMANN-PICK TYPE C (NPC)



WHAT IS NPC?

Niemann-Pick Type C is a rare lysosomal storage disease caused by inherited mutations in the NPC1 gene. This leads to a malfunction in the NPC1 protein, causing large molecules like cholesterol and lipids to accumulate in cells, especially in the brain, liver, and spleen. This damages these organs.

NPC symptoms are diverse and usually appear in childhood. They can include jaundice, an enlarged liver, ataxia (uncoordinated movement), and cognitive difficulties. Eventually, neurological decline proves fatal for people with NPC.

NPC affects 1 in 120,000 individuals. It can be caused by changes in two different genes: NPC1 accounts for 95% of cases and NPC2 accounts for 5%.

MANAGING SYMPTOMS WITH A REPURPOSED DRUG

N-acetyl-DL-leucine is a generic molecule used to treat vertigo.

Small US biotech IntraBio began by studying N-acetyl-DL-leucine as a treatment for a range of neurodegenerative diseases including NPC. They identified the active ingredient as the L enantiomer, N-acetyl-L-leucine (IB1001). They began formal clinical trials with IB1001

In June 2023, IntraBio released the results of a Phase III trial in which IB1001 reduced neurological signs and symptoms in NPC patients. They are now seeking a marketing authorisation for IB1001 for use in symptom management for NPC.

DESIGNING CLINICAL TRIALS

ONE DRUG, MANY POSSIBILITIES

The company is running multiple clinical trials across different conditions using one molecule. This maximises the chances of at least one group of patients benefiting from the research.

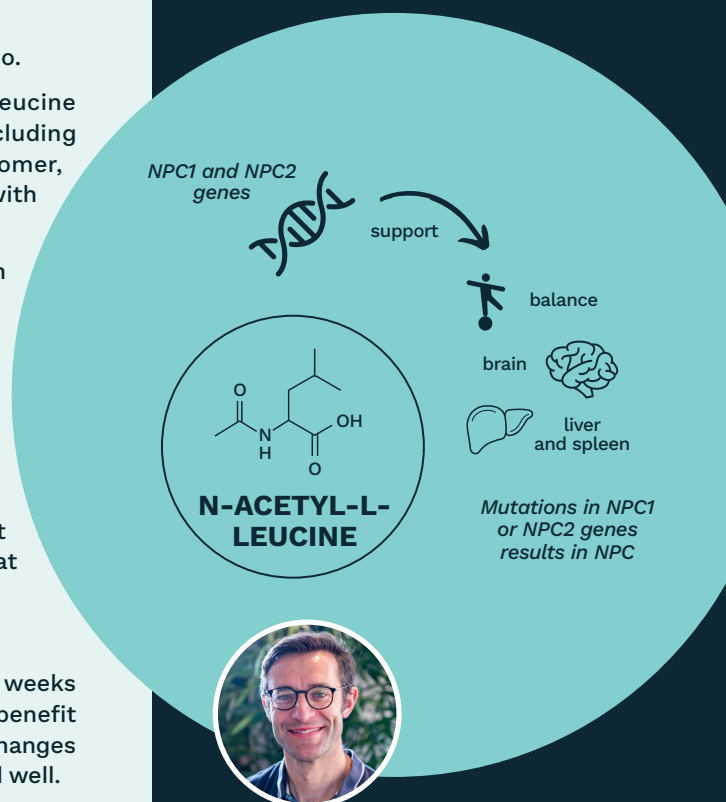
CROSS OVER TRIAL DESIGN

In this trial, participants took either the drug or placebo for 12 weeks and then swapped over. This meant everyone got a chance to benefit from treatment. This isn't appropriate for every indication, but changes in ataxia symptoms can be seen quickly so this design worked well.

AT A GLANCE

- NPC causes a range of symptoms, including neurological problems.
- Phase III trials shows N-Acetyl-L-Leucine is effective in treating a range of neurological symptoms in NPC patients.
- This drug is being explored for multiple indications.
- Understanding the underlying mechanisms helps researchers match diseases to treatments and get them to patients sooner.

WHAT IS NPC?



SPECIAL THANKS TO WILL EVANS

Will's eldest son, Sam, was diagnosed with NPC as an infant in 2008.

Will has been a trustee for Niemann-Pick UK since 2010. He chaired the charity between 2016 and 2022.



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