Treatment with N-acetyl-L-leucine beneficial for Niemann-Pick disease

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Treatment with N-acetyl-L-leucine (NALL) for 12 weeks yields better neurological status than placebo among patients with genetically confirmed Niemann-Pick disease type C, according to a study published in the Feb. 1 issue of the *New England Journal of Medicine*.

Tatiana Bremova-Ertl, M.D., Ph.D., from the University Hospital Bern in Switzerland, and colleagues randomly assigned 60 patients age 4 years or older with genetically confirmed Niemann-Pick disease type C to receive 12 weeks of NALL followed by 12 weeks of placebo or to receive 12 weeks of placebo followed by 12 weeks of NALL in a 1:1 ratio.

The researchers found that the mean change from baseline in the Scale for the Assessment and Rating of Ataxia total score was −1.97 ± 2.43 and −0.60 ± 2.39 points after 12 weeks of receiving NALL and placebo, respectively (least-squares mean difference, −1.28 points). For the secondary end points, the results were generally supportive of the findings in the primary analysis; however, these were not adjusted for multiple comparisons. A similar incidence of adverse events was seen with NALL and placebo; there were no treatment-related serious adverse events reported.

"In this phase 3 trial involving patients with Niemann-Pick disease type C, treatment with NALL reduced neurologic signs and symptoms as compared with placebo over 12 weeks," the authors write.

More information: Tatiana Bremova-Ertl et al, Trial of N-Acetyl-l-Leucine in Niemann–Pick Disease Type C, *New England Journal of Medicine* (2024). DOI: 10.1056/NEJMoa2310151

Cynthia J. Tifft et al, N-Acetyl-l-Leucine and Neurodegenerative Disease, *New England Journal of Medicine* (2024). DOI: 10.1056/NEJMe2313791