Scientists Discover Surprising Role of Vitamin B12 in Correcting Genetic Disorder – Related Defects

Overview

A common vitamin may hold the key to correcting developmental problems caused by a rare genetic disorder. Scientists have discovered that vitamin B12 can largely reverse growth and developmental abnormalities in laboratory worms carrying a mutation linked to Verheij syndrome.

The research, led by scientists at the Max Planck Institute for Biology of Ageing, investigated how genetic changes affecting RNA processing influence growth and metabolism.

Verheij syndrome is a rare genetic condition associated with developmental delays, poor growth and short stature.

Researchers focused on RNA splicing, a process that helps cells prepare genetic instructions needed to produce proteins.

When this process is disrupted by genetic mutations, normal cellular functions can be affected, potentially contributing to developmental disorders.

To investigate these effects, scientists used Caenorhabditis elegans, a tiny roundworm commonly used in medical research.

Worms carrying a mutation in an RNA-splicing gene associated with Verheij syndrome developed slowly and remained smaller than healthy worms.

Further experiments revealed that the mutation interfered with metabolic pathways dependent on vitamin B12, affecting processes important for normal growth.

Importantly, when researchers provided additional vitamin B12, the affected metabolic pathways returned towards normal function.

The treatment also largely corrected the developmental abnormalities observed in the worms.

Researchers subsequently examined human cell datasets and blood samples from patients diagnosed with Verheij syndrome.

These investigations revealed evidence of similar metabolic disturbances, suggesting that vitamin B12-related pathways may also be involved in the human condition.

However, the study did not establish that vitamin B12 supplementation improves symptoms in people with Verheij syndrome.

Researchers emphasised that findings from worm experiments cannot be directly applied to human treatment.

Further research is needed to determine whether targeting these pathways could eventually offer safe and effective treatments for affected patients.

REFERENCE: Jonathan Kölschbach, Hormos S. Dafsari, Emily Baum, Anna Löhrke, Chun Kew, Ivan Dikic, Wenming Huang, Adam Antebi. Vitamin B12 alleviates spliceosomopathy via phospholipid remodeling. Nature Communications, 2026; 17 (1) DOI: 10.1038/s41467-026-76295-9

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