

# First case of person without the protein needed to transport vitamin D identified

April 5 2019

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Dr. Julien Marcadier, MD, a clinical assistant professor in the departments of medical genetics and paediatrics at the Cumming School of Medicine and senior author of the paper. Photo by Kelly Johnston, Cumming School of Medicine.

Research identifying the first case of a person living without the protein needed to bind and transport vitamin D in the bloodstream has been published in the *New England Journal of Medicine*. The paper, led by a University of Calgary geneticist, and co-authored by researchers at the University of Washington in Seattle, U.S., and University of British Columbia (UBC), opens important new avenues of research into how the body transports and absorbs vitamin D, as well as how it can best be measured.

Vitamin D, a steroid hormone essential to [calcium regulation](#) and bone health, is available through a [healthy diet](#) or with supplements, and can also be made by the body when exposed to sunlight. Once in the bloodstream, most vitamin D becomes attached to the vitamin D binding protein for storage and transport. But researchers have identified a 59-year-old woman who lacks the binding protein and has no measurable vitamin D in her system—the first such human case.

"The idea has always been that without its binding protein, vitamin D couldn't get to the cells where it's needed. Our findings call that into question," says Dr. Julien Marcadier, MD, a clinical assistant professor in the departments of medical genetics and paediatrics at the Cumming School of Medicine and senior author of the paper. "To our surprise, she's maintained normal [calcium levels](#) despite having no measurable vitamin D. This tells us that the vitamin D she has in her system does in fact get to its target cells, though how exactly that happens is still unknown."

The woman's case puzzled doctors for years. In her early 40s, she started to break a few bones. "She had a number of fragility fractures, little falls where the average person wouldn't expect to break a bone," says Marcadier. Doctors found the woman had absolutely no vitamin D in her body, even after giving her massive doses. "No matter how much or how often they gave her vitamin D, the levels wouldn't budge above zero."

Dr. Patrick Ferreira, MD, now retired after years of service at the Alberta Children's Hospital, was the first geneticist brought in to help figure out the puzzle, and immediately suspected there was a problem with the woman's vitamin D binding protein. Marcadier then became involved, and was able to collaborate with teams at UBC and the University of Washington to confirm Ferreira's theory. The researchers proved that the woman is incapable of making any vitamin D binding protein because she lacks the gene to do so.

This opens up new avenues of research into how vitamin D and its binding protein work in the body, and how best to assess a person's vitamin D status. "Because her calcium and phosphate levels have been normal for years, we know that whatever vitamin D she is given can do its job," says Marcadier. "It also adds to some of the current controversy out there about how vitamin D should be measured, and what should be considered a normal level."

Animal research has shown that mice without the vitamin D binding protein can remain healthy, provided they have enough vitamin D in their diet. The same appears to be true for this unique patient.

This is the first reported case of a human without vitamin D binding [protein](#). The woman, who does not wish to be identified, is now taking vitamin D at a dose of 2,000 IU per day and has not suffered any recent fractures. She continues to work with her health-care team as they try to better understand the role of [vitamin D binding protein](#).

**More information:** Clark M. Henderson et al. Vitamin D–Binding Protein Deficiency and Homozygous Deletion of the GC Gene, *New England Journal of Medicine* (2019). [DOI: 10.1056/NEJMoa1807841](https://doi.org/10.1056/NEJMoa1807841)

Provided by University of Calgary

Citation: First case of person without the protein needed to transport vitamin D identified (2019, April 5) retrieved 19 April 2024 from <https://medicalxpress.com/news/2019-04-case-person-protein-vitamin-d.html>

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