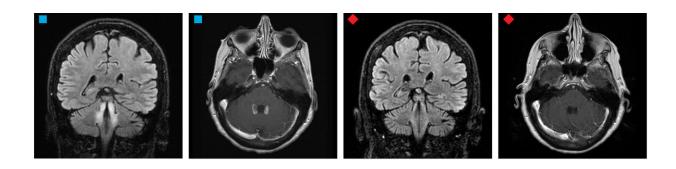


## Form of B12 deficiency in cerebral spinal fluid found to negatively affect the central nervous system

July 2 2024, by Bob Yirka



A patient with an unsual MRI and neurologic deficits eluded typical diagnostic tests. Credit: John Pluvinage

A large team of doctors and medical researchers with diverse backgrounds, affiliated with several institutions across the U.S., and led by a group at the University of California, San Francisco, has found what might be a previously unknown kind of autoimmune disease—one that negatively impacts the central nervous system.

In their paper published in *Science Translational Medicine*, the group describes how they found a form of B12 deficiency in a patient who had normal levels of the vitamin in their blood, and what they learned as they looked deeper.



In 2014, a team of doctors found themselves having difficulty diagnosing a 67-year-old female patient with symptoms typically associated with B12 deficiency—she had difficulty speaking, tremors and ataxia. Blood tests showed no B12 deficiency.

Eventually, the patient was enrolled in a study that had researchers looking into patients with novel autoantibody ailments—most of those in the study, including the original patient, had signs of neuroinflammatory disease.

The team conducted a <u>spinal tap</u> to analyze the patients' cerebral spinal fluid. What came back was a surprise—the patient had almost no B12 in the fluid that bathes the brain and <u>central nervous system</u>, suggesting something was preventing it from crossing the blood brain barrier.

In their search for an explanation, the researchers discovered that the patient had autoantibodies that were targeting her CD320 receptors—molecules on the cells that normally help carry B12 across the <u>blood brain barrier</u>.

After giving the patient an immunosuppressant and large doses of B12 supplements, they found her symptoms subsiding and her levels of B12 in cerebral spinal fluid increasing.

Surprised by their findings, the research team tested 254 other patients who were enrolled in the same study and found that seven had the same type of autoantibodies and four had normal levels of B12 in the blood, but very low levels in their cerebral spinal fluid. They also found what might be an association with lupus.

The team concludes by suggesting that their findings indicate that they may have uncovered a previously unknown kind of autoimmune disease, one with "varied neurologic manifestations."



**More information:** John V. Pluvinage et al, Transcobalamin receptor antibodies in autoimmune vitamin B12 central deficiency, *Science Translational Medicine* (2024). DOI: 10.1126/scitranslmed.adl3758

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