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Cobalamin defects can explain neurologic regression in children

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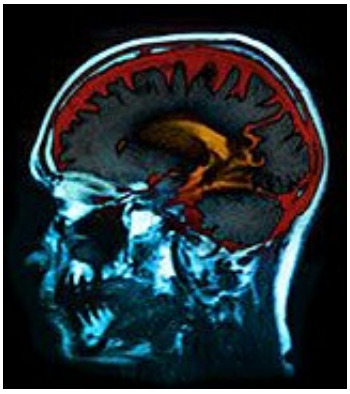

(HealthDay)—Cobalamin defects can account for neurologic regression in healthy children, according to a case report published online Dec. 15 in *Pediatrics*.

Jena M. Krueger, M.D., from the Northwestern University Feinberg School of Medicine in Chicago, and colleagues reported the case of an 8-year-old boy who presented with a month-long history of waxing and waning encephalopathy and acute regression followed by seizures.

Following evaluation for a metabolic disorder, the researchers identified methylmalonic acidemia and hyperhomocysteinemia of the cobalamin C type, which was a result of a single, presumed homozygous pathogenic

c.394 C>T mutation in the *MMACHC* gene. The patient improved significantly with vitamin replacement and appropriate diet restrictions, and returned to his premorbid level of behavior.

"This case illustrates an unusual presentation of a treatable metabolic disorder and highlights the need to consider cobalamin defects in the differential diagnosis of healthy children with neurologic regression," the authors write.

More information: Abstract
Full Text (subscription or payment may be required)