

Clinical signs of citrin deficiency mimic anorexia nervosa

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The clinical features of citrin deficiency may mimic those of anorexia nervosa, according to a case report published online July 20 in *Pediatrics*.

(HealthDay)—The clinical features of citrin deficiency (CD) may mimic those of anorexia nervosa (AN), according to a case report published online July 20 in *Pediatrics*.

Satsuki Takeuchi, M.D., from the Matsumoto Medical Center of Chushin-Matsumoto Hospital in Japan, and colleagues reported the case of a 12-year-old female presenting with severe anorexia and weight loss, mimicking the restricting type of AN. The patient showed normal development up to age 10 when she started to play volleyball at school. Thereafter, she became anorexic, and her growth was stunted. She was admitted to the hospital with severe anorexia and thinness at age 12.



The patient was initially thought to have AN and was given a drip infusion of glucose solution and high-calorie drinks, but her condition declined further. Having a history of neonatal hepatitis, she was suspected of having a CD. A diagnosis was made after genetic analysis of *SLC25A13*, which showed that she was compound heterozygous for 851del4 and IVS16ins3kb. She was started on a low-carbohydrate diet with oral intake of arginine and ursodeoxycholic acid; gradual improvement was seen in her condition.

"The clinical features in our patient were similar to those of AN, and therefore AN may also be an important clinical sign in adolescent patients with CD," the authors write.

More information: Abstract

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