

# Abstract

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## **A case of partial biotinidase deficiency associated with autism.**

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**BACKGROUND:** We report the case of a child with partial biotinidase deficiency and autistic developmental disorder. We arrived at the diagnosis of biotinidase deficiency when the child was almost 4 years of age.

**DISCUSSION:** Consequently, he began cofactor biotin treatment (10 mg daily) which did not resolve his autistic behavior. His younger brother was affected by partial biotinidase deficiency diagnosed at birth through our neonatal screening program. He was precociously treated with cofactor biotin therapy (10 mg daily) and did not show any behavioral abnormality or developmental delay. **Since the brain is quite vulnerable to biotin deficiency, delayed biotin therapy could result in neurological damage. Our patient is the first case of partial biotinidase deficiency associated with autism.**

**CONCLUSION:** We hypothesize that the low biotinidase activity could have caused biotin deficiency in his brain and cerebrospinal fluids and consequently serious neurological problems, such as stereotyped and autistic behaviors, which were irreversible in spite of biotin supplementation.

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