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Letter to the Editor

Any individual with multiple sclerosis who markedly improves neurologically with high-doses of biotin should be evaluated for biotinidase deficiency

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Barry Wolf

Couloume and colleagues[1] have provided further evidence that very high doses of biotin do not improve the vast majority of individuals with progressive multiple sclerosis in a large cohort. These findings support those of a previous study of 43 individuals with the disorder who, for the most part, also did not improve on enormous doses of biotin.[2] However, in Couloume et al.'s study, of the 178 individuals treated with biotin six exhibited some improvement.

There have been an increasing number of reports of individuals who were initially presumed to have multiple sclerosis or related diseases who actually had profound biotinidase deficiency (less than 10% of mean normal serum biotinidase activity), an autosomal recessively inherited disorder that is treatable with pharmacological doses of biotin (10–20 mg per day).[3–5] These adults with biotinidase deficiency usually present with peripheral neuropathies with or without optic neuropathy, symptoms that are different from those of children with disorder. This is a major reason why biotinidase deficiency is not usually included in the differential diagnosis of individuals with neuropathies. It is important to make the correct diagnosis of biotinidase deficiency rapidly and begin treatment before the symptoms become irreversible. Enzymatic testing in serum is conclusive and inexpensive. If an individual is shown to have the enzyme deficiency, they would only require a pharmacological dose of biotin for treatment, rather than the megadoses of biotin currently recommended for individuals with multiple sclerosis. In addition, we still do not know the possible side effects of such enormous doses of biotin recommended for those with progressive multiple sclerosis.

Because biotinidase deficiency is readily treatable and is not usually included in the differential diagnosis of individuals with multiple sclerosis, I suggested in a letter to the editor[6] that testing for biotinidase deficiency should be performed in all individuals suspected as having multiple sclerosis.[7] However, this recommendation may not be considered practical or heeded routinely. Therefore, the small group of individuals that Dr Couloume and his colleagues found who improved with biotin therapy may be candidates for biotinidase deficiency testing. However, any individuals thought to have multiple sclerosis and who exhibit optic neuropathy and/or myelopathy that improves markedly with biotin therapy should be evaluated for biotinidase deficiency.

Correspondence to:
Barry Wolf,
Attending Physician in the Division of Genetics, Birth Defects and Metabolic Diseases of the Department of Pediatrics, Ann and Robert H. Lurie Children's Hospital of Chicago and Professor of Pediatrics, Northwestern Feinberg School of Medicine, 225 E. Chicago Ave., Chicago, IL 60611, United States of America.
barrywolf1@comcast.net

Barry Wolf,
Division of Genetics, Birth Defects and Metabolic Diseases of the Department of Pediatrics, Ann and Robert H. Lurie Children's Hospital of Chicago and Professor of Pediatrics, USA

ORCID iD

Barry Wolf https://orcid.org/0000-0003-2893-4983

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