

disease [3]. Inheritance of the APOE e4 genotype is an independent risk factor for developing higher levels of amyloid accumulation.

References.

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2. Growdon JH, Hyman BT. JAMA Neurol. 2014 Jan 1;71(1):7-8.
3. Hyman BT, et al. Arch Neurol. 1995 Apr;52(4):373-8.

RIBOFLAVIN IN BROWN-VIALETTO-VAN LAERE SYNDROME

Investigators at Great Ormond Street Hospital, London, UK, and multiple centers internationally report the response to high-dose oral riboflavin therapy in 18 patients from 13 families with mutations in SLC5ZA2, encoding riboflavin transporter RTVT2, a new causative gene for Brown-Vialetto-Van Laere syndrome (BVVLS), a progressive neurodegenerative disorder leading to death in childhood. BVVLS is characterized by cranial neuropathies, pontobulbar palsy, sensorimotor neuropathy manifesting with sensory ataxia, weakness of upper limbs and axial muscles, with preserved strength of lower limbs, optic atrophy, sensorineural hearing loss, and respiratory insufficiency. Riboflavin therapy resulted in significant sustained clinical and biochemical improvement in 2 patients and preliminary response in 13 patients. (Foley AR, Menezes MP, Pandraud A, et al. Treatable childhood neuronopathy caused by mutations in riboflavin transporter RFVT2. **Brain** 2014 Jan;137(Pt 1):44-56).

COMMENTARY. BVVLS is a similar disorder to Fazio Londe syndrome caused by subtly different mutations of the same gene, and with the additional clinical feature of sensorineural deafness [1][2]. Diagnosis requires mutation analysis of transporter genes. The simple treatment with riboflavin supplementation may halt progression of both neurodegenerative disorders. An invited comment by Dr. John Wilson, Emeritus Chief of Neurology, Great Ormond Street Hospital, London, UK, and an authority on Fazio-Londe disease [2], is paraphrased as follows: “as our understanding of the basic concepts of disease become more complex, so we are lead to a beautiful simplicity (in the form of vitamin therapy) that brings light into dark places.” How many similar degenerative diseases may in the future be found responsive to a simple vitamin?

References.

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2. McShane MA, et al. Brain. 1992 Dec;115 (Pt 6):1889-900.

AUTISM SPECTRUM DISORDERS

ABNORMAL MOTOR FUNCTION AND AUTISM

Investigators from Albert Einstein College of Medicine, Bronx, NY, recorded the gait characteristics and prevalence of toe walking, the range of passive joint mobility, and age at walking in children with DSM IV autism spectrum disorders (ASDs) and in age- and gender-matched healthy peers (mean age 4 years 6 months, range 22 months – 10