Idiopathic intracranial hypertension in cystinosis.

Dogulu CF, Tsilou E, Rubin B, Fitzgibbon EJ, Kaiser-Kupper MI, Rennert OM, Gahl WA.

Laboratory of Clinical Genomics, National Institute of Child Health and Development, Ophthalmic Clinical Genetics Section, National Institutes of Health, Bethesda, MD 20892-4429, USA. doguluc@mail.nih.gov

Abstract

OBJECTIVES: To report a high frequency of idiopathic intracranial hypertension (IIH) in patients with cystinosis and to speculate on the relationship between these two disorders. STUDY DESIGN: Retrospective case series and review of the literature regarding risk factors for the development of IIH in cystinosis. RESULTS: Eight patients with cystinosis had documented papilledema, normal neuroimaging of the brain, cerebrospinal fluid (CSF) opening pressure greater than 200 mm of H2O, and normal CSF composition. No common medication, condition, or disease except cystinosis was found in these persons. Six of the patients had received prednisone, growth hormone, cyclosporine, oral contraceptives, vitamin D, or levothyroxine at the time of onset of IIH. Five patients had previous renal transplants. CONCLUSION: No single risk factor for the development of IIH linked IIH to cystinosis in our patients. However, thrombosis susceptibility as a result of renal disease or impaired CSF reabsorption in the arachnoid villi as a result of cystine deposition might lead to the development of IIH in cystinosis.