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effective October 1st, 2016

Journal of Inherited Metabolic Disease

— Official Journal of the Society for the Study of Inborn Errors of Metabolism



Profile The **JIMD** aims to improve the management and understanding of inherited metabolic disorders by enhancing communication between scientists in the field throughout the world. It publishes results of original research and new or important observations pertaining to any aspect of inherited metabolic disease in humans and higher animals. This includes clinical (medical, dental and veterinary), biochemical, genetic (including cytogenetic, molecular and population genetic), experimental (including cell biological), methodological, theoretical, epidemiological, ethical and counselling aspects.

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Affiliation » Society for the Study of Inborn Errors of Metabolism, SSIEM.

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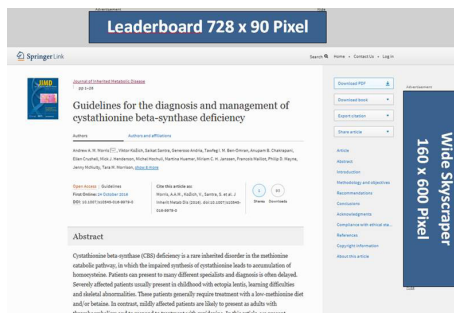
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Guidelines for the diagnosis and management of cystathionine beta-synthase deficiency

Authors: Address and affiliation: Andrew A. H. Morley, Hans-Hilbert, Sabina Sauer, Gersona de la Torre, Ulf Ben-David, Alexander B. Chapman, Hans-Cristian, et al. | Download PDF | Download image | Export citation | Share article

Abstract: Cystathionine beta-synthase (CBS) deficiency is a rare inherited disorder in the methionine metabolic pathway, in which the impaired synthesis of cystathionine leads to accumulation of homocysteine. Patients can present to many different specialists and diagnosis is often delayed. Severely affected patients usually present in childhood with ataxia, keratitis, hearing difficulties and skeletal abnormalities. These patients generally require treatment with a low-methionine diet and/or betaine. In contrast, mildly affected patients are likely to present as adults with thrombotic/vascular and/or renal problems. In this article, we present

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