

Sandhoff Disease—A Case Report of 3 Siblings and a Review of Potential Therapies

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Abstract

Introduction: Sandhoff disease is a G_{M2} gangliosidosis that may present within the first 6 months of life with developmental regression. This is the first report of a pedigree from Southeast Asia. **Clinical Picture:** All the affected siblings presented in the first year of life with developmental regression, spasticity, seizures and loss of vision. The diagnosis was confirmed by an enzymatic deficiency in both β -hexosaminidase A and B. **Conclusion:** As the disorder is autosomal recessive, and no curative therapy is currently available, genetic counselling is necessary to prevent the burden of this devastating disease. We review the potential strategies of treatment for Sandhoff disease.

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Key words: Autosomal recessive, β -hexosaminidase, Bone marrow transplant, Developmental regression, G_{M2} gangliosidosis

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