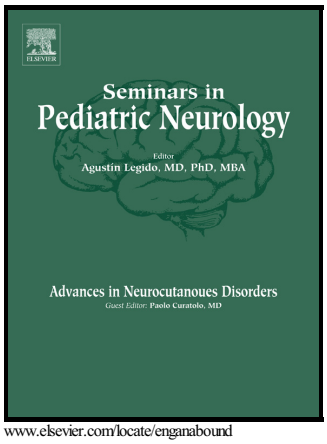

The first Case of Riboflavin Transporter Deficiency in Sub-Saharan Africa

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The first case of riboflavin transporter deficiency in sub-Saharan Africa

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Abstract

This report describes the first case of a child with genetically confirmed Brown-Vialetto-Van Laere syndrome (BVVL) in sub-Saharan Africa. This is an extremely rare clinical condition that presents with an auditory neuropathy, bulbar palsy, stridor, muscle weakness and respiratory compromise that manifests with diaphragmatic and vocal cord paralysis. It is an