Consideration of genetic diagnoses of developmental delay in children of consanguineous families

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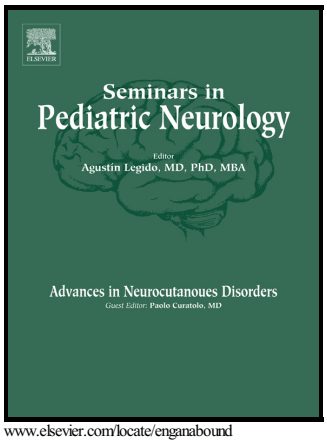

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Title

Consideration of Genetic Diagnoses of Developmental Delay in Children of Consanguineous Families

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Abstract

In patients presenting with global developmental day, eliciting a history of consanguinity may increase a clinician's bias towards suspecting an autosomal recessive aetiology. We present 3 cases wherein children of consanguineous parents presented to the pediatric neurology clinic for evaluation and potential diagnosis of the cause of global developmental delay. The outcome of the investigations in each case demonstrate the need to follow established guidelines for appropriate genetic testing as they pertain to the patient's presentation rather than a single element of the history (ie. consanguinity).