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Joubert Syndrome in Children as Multicentric Screening in Ten Different Places in World

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Abstract : Introduction: Joubert syndrome has an autosomal recessive pattern of inheritance. It is referred as the brain malfunctioning and caused due to the underdevelopment of the cerebellar vermis. Associated conditions involving the eye, the kidney, and ocular disease are well described. Aims: Research helps us better understand this diseases, Joubert syndrome and can lead to advances in diagnosis and treatment. Methods: Different several conditions have been described in which the molar tooth sign and characteristics of Joubert syndrome in ten different places in the world. Carrier testing and diagnosis are available if one of these gene mutations has been identified in an affected family member. Results: Authors have described eleven cases during twenty years of Joubert syndrome. It is a clinically and genetically heterogeneous group of disorders characterized by hypoplasia of the cerebellar vermis with the characteristic neuroradiologic molar tooth sign, and accompanying neurologic symptoms, including dysregulation of breathing pattern and developmental delay. We made confirmation of diagnosis in twin sisters with Joubert syndrome with renal anomalies. Ocular symptoms have existed in seven cases (63.64%) from total eleven. Eleven cases were different sex, five boys (45.45%) and six girls (54.44%). Conclusions: Joubert syndrome is inherited as an autosomal recessive genetic disorder with several features of the disease.

Keywords : Joubert syndrome, cerebellooculorenal syndrome, autosomal recessive genetic disorder (ARGD), children
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