

Joubert Syndrome and Related Disorders: A Single Center Experience

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Abstract : Background and objective: Joubert syndrome (JS) is a rare, autosomal-recessive condition. Early recognition is important for management and counseling. Magnetic resonance imaging (MRI) can help in diagnosis. Therefore, we sought to evaluate clinical presentation and MRI findings in Joubert syndrome and related disorders. Method: A retrospective review of genetically proven cases of Joubert syndromes and related disorders was reviewed for their clinical presentation, demographic information, and magnetic resonance imaging findings in a period of the last 10 years. Two radiologists documented magnetic resonance imaging (MRI) findings. The presence of hypoplasia of the cerebellar vermis with hypoplasia of the superior cerebellar peduncle resembling the “Molar Tooth Sign” in the mid-brain was documented. Genetic testing results were collected to label genes linked to the diagnoses. Results: Out of 12 genetically proven JS cases, most were females (9/12), and nearly all presented with hypotonia, ataxia, developmental delay, intellectual impairment, and speech disorders. 5/12 children presented at age of 1 or below. The molar tooth sign was seen in 10/12 cases. Two cases were associated with other brain findings. Most of the cases were found associated with consanguineous marriage Conclusion and discussion: The molar tooth sign is a frequent and reliable sign of JS and related disorders. Genes related to defective cilia result in malfunctioning in the retina, renal tubule, and neural cell migration, thus producing heterogeneous syndrome complexes known as “ciliopathies.” Other ciliopathies like Senior-Loken syndrome, Bardet Biedl syndrome, and isolated nephronophthisis must be considered as the differential diagnosis of JS. The main imaging findings are the partial or complete absence of the cerebellar vermis, hypoplastic cerebellar peduncles (giving MTS), and (bat-wing appearance) fourth ventricular deformity. Limitations Single-center, small sample size, and retrospective nature of the study were a few of the study limitations.

Keywords : Joubert syndrome, magnetic resonance imaging, molar tooth sign, hypotonia

Conference Title : ICR 2022 : International Conference on Radiology

Conference Location : Dubai, United Arab Emirates

Conference Dates : December 20-21, 2022