A 35 year old female presented in the emergency room with history of focal seizures involving the right side of the face. There was also history of nystagmoid eye movements during the episode. There was history of similar episodes of seizures since childhood. There was no history of any comorbid illness. Patient had mild mental retardation since childhood. She was born of a non-consanguineous marriage with no family history of any seizure disorder. On examination, the patient was conscious, oriented, afebrile, no neurocutaneous markers were found. Intelligent Quotient (IQ) was 70. Pupil 3mm equally reacting to light on both sides. Fundus examination was normal. Plantars were flexor. On investigating the patient, the hemogram, renal and liver function tests were normal. Random blood glucose was 120mg/dl. Ultrasonogram of abdomen and pelvis was normal. MRI (T2 Axial) also showed Bat wing appearance of 4th Ventricle (Figure 2). EEG was normal. Diagnosis of Joubert syndrome was made.

Joubert syndrome (JS) was originally described in 1968 in four siblings with agenesis of the cerebellar vermis presenting with episodic hyperpnoea, abnormal eye movements, ataxia and intellectual disability.1 Several years later, a pathognomonic midbrain-hindbrain malformation, the “molar tooth sign” (MTS), was detected first in JS,2 and then in several other conditions previously considered as distinct entities. The term “Joubert syndrome and Related Disorders” (JSRD) was then coined to group all conditions sharing abnormalities of respiratory pattern. The diagnosis of JSRD is suspected in patients presenting with hypotonia, oculomotor apraxia, nystagmus and abnormalities of respiratory pattern. The management is essentially symptomatic with antiepileptics.

This case highlights the rare association of Joubert syndrome with seizures presenting with a typical radiological sign on imaging study.

References


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Fig. 1: MRI Brain (T2 Axial) showing typical molar tooth sign

Fig. 2: MRI Brain (T2 Axial) showing bat wing appearance of fourth ventricle