JOUBERT SYNDROME: A RARE AUTOSOMAL RECESSIVE DISORDER

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ABSTRACT

Joubert syndrome is an extremely rare autosomal recessive disorder. It is diagnosed primarily on the basis of imaging and clinical findings. Generally diagnosed in infancy, the cases present with a constellation of signs, mainly cerebellar. Imaging by MRI (Magnetic Resonance Imaging) reveals the pathognomonic 'molar tooth' sign and 'bat wing' sign. At our institution, a 7 month old infant presented with abnormal breathing and eye movements. He was examined and found to have hypotonia and delayed development, in addition to nystagmus and hyperpnoea. On MRI scanning he revealed the typical imaging findings, prompting us to report this rare case.

Keywords: Joubert syndrome, molar tooth, bat wing, MRI

Case Report

A 7 month old infant presented to us with nystagmus, hyperpnoea, hypotonia and delayed developmental milestones. The infant underwent MRI examination of brain, which revealed the characteristic molar tooth sign in axial sequences (caused by inadequate decussation of fibers in the superior cerebellar peduncles, leading to abnormality in their appearance and hence the sign (Fig. 1). The bat wing sign was demonstrated as well (this sign occurs due to hypoplasia of the vermis, which leads to the formation of a midline cleft between the cerebellar hemispheres and consequently the bat wing appearance of the fourth ventricle on axial MRI sequences) (Fig. 2).

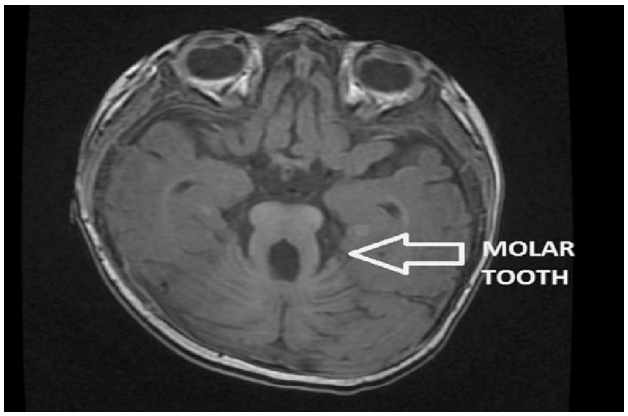

Figure 1: Axial T1W image at the level of the midbrain showing molar tooth appearance to horizontally disposed dysplastic cerebellar peduncles.

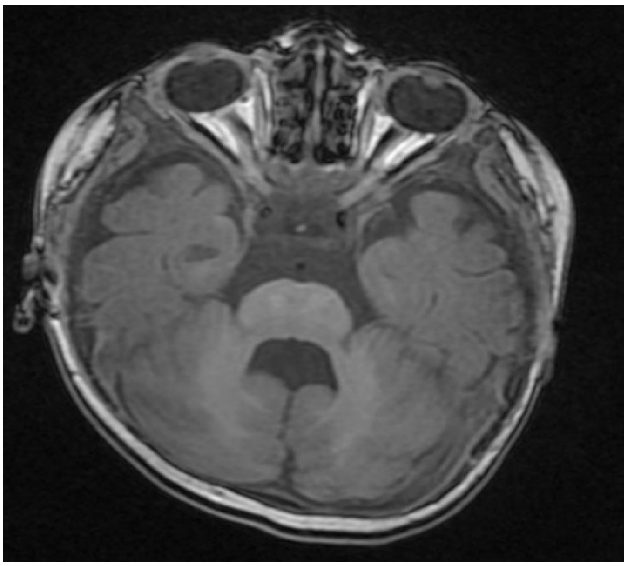

Figure 2: Axial T1W at the level of ponto-medullary junction showing bat wing appearance of fourth ventricle. The cerebellar hemispheres are nearly apposed due to the hypoplastic vermis.

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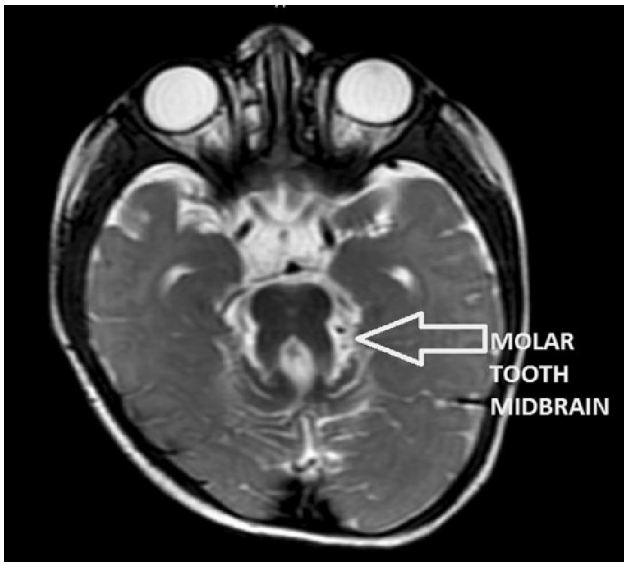

Figure 3: Axial T2W image showing molar tooth appearance.

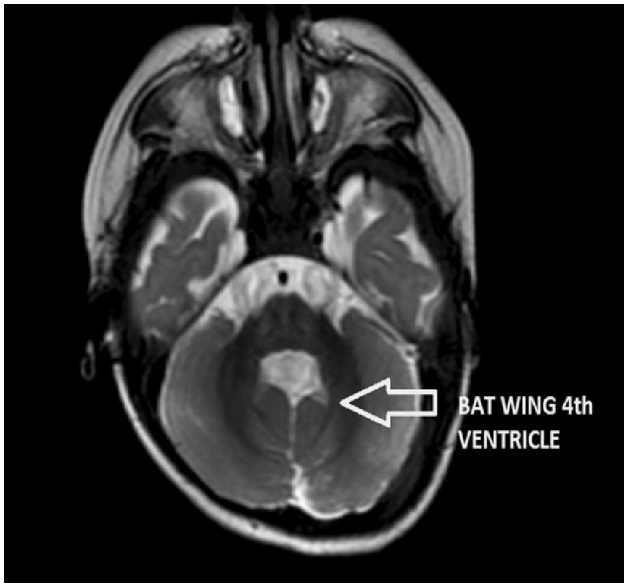

Figure 4: Axial T2W image showing bat wing

Discussion

Joubert syndrome was first described by French neurologist Marie Joubert in 1969.[4] Clinical features of this syndrome consist of nytstagmus, hypotonia, hyperpnoea and delayed motor development.[1-3] The main pathology being aplasia or hypoplasia of vermis and abnormality of the pontomesencephalic junction. Other associated findings are retinal coloboma, multicystic kidney disease, tongue hamartoma and polydactyly.[5,6] Kidney involvement generally occurs in association with retinal involvement.

Imaging findings are characteristic. There is hypoplasia or aplasia of the cerebellar vermis and thickened, horizontally disposed cerebellar peduncles (leading to the molar tooth sign). Both these features result in a deformed fourth ventricle (hence causing the bat wing morphology of the fourth ventricle).

Usually no supratentorial abnormalities are seen. However, few cases have been reported to have corpus callosal dysgenesis[7] and enlarged lateral ventricles.

Conclusion

Joubert syndrome is a rare neurological disorder where modern imaging has immensely helped in clinching the diagnosis.It has been infrequently reported in literature, thus prompting us to report this case, diagnosed at our institution.

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