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Radiology

"Molar Tooth" Sign Pathognomonic of Joubert Syndrome

Aloumba-Gilius Donald Wilhem[1*], Ali Mheimed[1], Johane Dzota[1], Daoud Bentaleb[1], Dalale Laouidiyi[1], Kamilia Chbani[1], Siham Salam[1]

[1]Radiology Department, Mother and Child Hospital Abderrahim Harouchi, University Hospital Ibn Rochd Casablanca, Morocco

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***Corresponding author:** Aloumba-Gilius Donald Wilhem
Radiology Department, Mother and Child Hospital Abderrahim Harouchi, University Hospital Ibn Rochd Casablanca, Morocco

Abstract **Original Research Article**

Joubert syndrome is a rare autosomal disorder characterised by abnormal respiratory patterns, delayed psychomotor development, visual disturbances and renal and hepatic involvement. We report the case of a 7-year-old girl who presented with delayed psychomotor development. Neurological examination revealed hypotonia. No respiratory disorders were observed in our case. Magnetic resonance imaging revealed the pathognomonic sign of the molar tooth in the posterior cerebral fossa.

Keywords: Joubert syndrome, visual disturbances, molar tooth, psychomotor development.

INTRODUCTION

Joubert syndrome is a rare genetic disorder of autosomal recessive inheritance affecting the cerebellum and brainstem. Its prevalence is 1 in 100,000 children [1]. It is manifested in the neonatal period by irregular breathing and nystagmus, and in childhood by cerebellar ataxia, developmental delay, oculomotor apraxia and/or convulsions. Facial and ocular abnormalities are also possible.

Objectif

Using a paediatric observation, to show the appearance of Joubert syndrome on magnetic resonance imaging (MRI).

MATERIALS AND METHODS

The case involved a 7-year-old girl from a second-degree consanguineous marriage, who was being monitored for delayed psychomotor development. Clinical examination revealed hypotonia, and a cerebral MRI was performed at the radiology department of the Abderrahim Harouchi hospital in Casablanca.

Résultats

Cerebral MRI revealed: a deeper interpeduncular fossa associated with hypoplasia of the cerebellar vermis, as evidenced by horizontalization of the upper cerebellar peduncles in sagittal reconstruction, producing the "molar tooth" sign (Fig 1); associated with this was a right retro-cerebellar cystic formation pushing V4 upwards and responsible for scalloping on the occiput. Joubert syndrome was diagnosed on the basis of clinical and MRI findings.

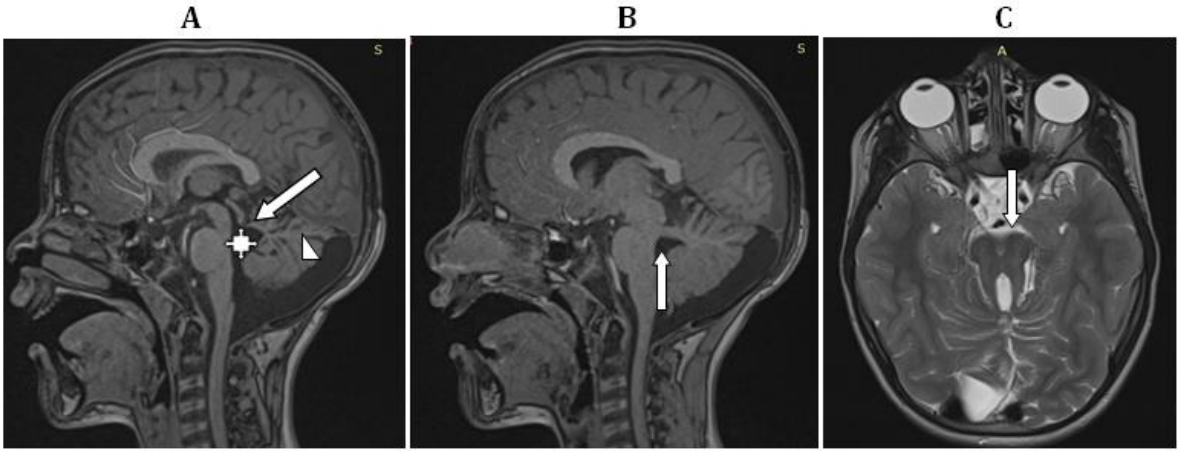

Figure 1: Brain MRI: A) T1 median sagittal section of the posterior cerebral fossa: moderate hypoplasia of the cerebellar vermis (arrowhead), dilation and opening of the V4 (asterisk), deepened interpeduncular fossa (arrow). B) T1 paramedian sagittal section of the posterior cerebral fossa showing thickening and horizontalization of the superior cerebellar peduncle (arrow). C) T2 axial section of the posterior cerebral fossa showing thickened and horizontal cerebellar peduncles with the characteristic "molar tooth" appearance (arrow)

DISCUSSION

The pathognomonic sign on MRI is that of the molar tooth, which guides the radiological diagnosis of Joubert syndrome. It results from a deep interpeduncular fossa, thick and elongated upper cerebellar peduncles, and hypoplasia of the cerebellar vermis [2]. The umbrella sign may be observed due to hypoplasia of the cerebellar vermis responsible for dilatation of the fourth ventricle. Other signs may include abnormalities of the corpus callosum, cerebellum, hippocampus or cerebral cortex [3, 4]. Ultrasound may reveal associated malformations (renal cysts and hepatic fibrosis and cysts). The differential diagnosis is made with other malformations of the posterior fossa but without a sign of the molar tooth: for agenesis of the vermis (Dandy Walker malformation, Rhombo-encephalo-synapsis), for cerebellar hypoplasia (pontone-cerebellar hypoplasia) and focal or diffuse dysplasia [5]. Management is symptomatic and multidisciplinary.

CONCLUSION

Joubert syndrome is a rare, inherited, autosomal recessive syndrome characterised by neonatal respiratory disorders, abnormal eyeball movements and later onset of psychomotor retardation with hypotonia and oculomotor apraxia. Comparison of clinical data with characteristic MRI images (molar tooth sign) enables a positive diagnosis to be made, which is the only guarantee of genetic counselling and antenatal diagnosis.

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